**Genetics Society of Australasia 2007** 54<sup>th</sup> Annual Conference, June 26<sup>th</sup>-29th The University of Sydney, Australia

**Comparative Genomics Workshop June 26th -27th** 







# ABSTRACTS



**GSA 2007** 

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**Genetics Society of Australasia 2007** 54<sup>th</sup> Annual Conference, June 26<sup>th</sup>-29th The University of Sydney, Australia

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# 54<sup>th</sup> Annual Meeting of Genetics Society of AustralAsia, Inc 26<sup>th</sup> June to 29<sup>th</sup> June, 2007

and

# **Comparative Genomics Workshop**

26<sup>th</sup> June and 27<sup>th</sup> June, 2007

# at University of Sydney

# **CONFERENCE ABSTRACTS**

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Genetics Society of AustralAsia, Inc c/o Department of Genetics University of Melbourne Victoria 3010, Australia

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# **Conference Organising Committee**

Kathy Belov (Convenor) Sonia Cattley Janine Deakin Mark Eldridge Karen Firestone Rebecca Johnson Nathan Lo Chris Moran Denis O'Meally Matthew Wakefield Alan Wilton

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# **Plenary speakers**

- Richard Gibbs (Baylor College of Medicine)
- Scott Edwards (Harvard University)
- Dave Burt (Roslin Institute)
- Win Hide (University of the Western Cape)
- Marilyn Raymond (National Institutes of Health)
- Ruth Hall (University of Sydney)
- Alan Cooper (University of Adelaide)
- Jeremy Timmis (University of Adelaide)
- Richard Harvey (Victor Chang Institute)
- John Mattick (University of Queensland)
- Scott O'Neill (University of Queensland)
- Barry Brook (University of Adelaide)
- Marilyn Renfree (University of Melbourne)
- Bill Ballard (University of New South Wales)

### Richard A Gibbs (Baylor College of Medicine)

Professor Richard Gibbs is Director of the Human Genome Sequencing Center, Baylor College of Medicine. After developing technologies for rapid genetic analysis early in his career, he became a key player in human genome project, and has overseen numerous recent projects, including the rat genome and human chromosomes 3 and 12.

### Scott Edwards (Harvard University)

Professor Scott Edwards holds a chair in Organismic and Evolutionary Biology at Harvard University, and is Curator of Ornithology in the Museum of Comparative Zoology. His current major interests include genome and sex chromosome evolution and phylogenetics in amniotes using BAC libraries and other resources; speciation analysis and historical demography using multilocus SNP loci; estimation of recombination rates and linkage disequilibrium in natural populations; behavioral and ecological consequences of MHC variation; and QTL mapping in passerine birds.

#### Dave W Burt (Roslin Institute)

Professor David Burt is a research group leader at the Roslin Institute (Edinburgh), Director of the ARK-Genomics facility (www.ark-genomics.org) and is the co-coordinator of AvianNET (www.chicken-genome.org) the international chicken genome network. He graduated in Molecular Biology from Edinburgh University in 1978; studied the molecular genetics of bacteriophage lambda at Leicester University and received his PhD in 1980. He was a research associate at the ICI/University of Leicester Joint Laboratory from 1980-1985, Harvard Medical School from 1985-1987, and Medical Research Council from 1987-1988, involved mainly in structural and gene expression studies in bacteria, rodents and humans. In 1988, he was appointed as a principal investigator at the Roslin Institute where he now leads national and international research on the genomics of the chicken and other birds.

#### Win Hide (University of the Western Cape)

Professor Winston Hide is Chair of Genomics and Bioinformatics at The University of the Western Cape and Director of the South African National Bioinformatics Institute. He focuses his research in two areas: unraveling the gene deregulation processes which initiate the development of cancers, and determination of the interactions of genes that protect the body from pathogens such as HIV. Hide has developed broadly used algorithms and systems for organisation of gene expression information, including d2, STACKdb, StackPack and eVOC. Hide's major interest in development of health solutions in South Africa revolves around the establishment of a cadre of international quality health science professionals, able to lead in the management and development of solutions for epidemic problems in Africa.

### Marilyn Raymond (National Institutes of Health)

Dr. Menotti-Raymond is a staff scientist at the National Cancer Institute-Frederick, Frederick, MD. Her research has focused on the generation of genetic maps in the domestic cat in order to characterize genes associated with hereditary disease and related biological interest. Dr. Raymond's group has recently mapped and characterized a mutation causative of late onset retinal atrophy in the Abyssinian cat, a model of human retinitis pigmentosa, and identified the causative mutation in a unique gene (LIX1) for spinal muscular atrophy. They have mapped and characterized mutations associated with coat color (chocolate, cinnamon, Burmese, Siamese) and the dilution of coat color (dilute) in the cat. Additionally, Dr. Raymond has been involved with application of genetic markers in the cat for forensic analysis, which led to the first introduction of an animal DNA fingerprint into court and the development of an STR genotyping system for genetic individualization of cat specimens.

### Ruth M Hall (University of Sydney)

Professor Ruth Hall is Senior Principal Research Scientist at the School of Molecular and Microbial Biosciences at The University of Sydney. Well-known for her discovery of integrons, she investigates the mobility of DNA in bacteria, particularly where it is important in the dissemination of antibiotic resistance genes and the evolution of new pathogens. Her focus is on identifying and unravelling new mechanisms for gene movement that are important in the dissemination of resistance genes. She also studies the biochemistry of these processes.

### Alan Cooper (University of Adelaide)

Professor Alan Cooper is Australian Research Council Federation Fellow and Director of the Australian Centre for Ancient DNA, The University of Adelaide. Cooper specialises in using ancient DNA to record and study evolutionary processes in real time, especially those associated with environmental change. His work ranges over timescales of hundreds of years old to permafrost-preserved bones of mammals and sediment dating to >300 kyr. His research is characterised by multi-disciplinary approaches involving the combination of information from areas such as geology, archaeology, anthropology, and even forensics to provide novel views of evolution, population genetics and palaeoecology.

### Jeremy N. Timmis (University of Adelaide)

Professor Jeremy Timmis is in the discipline of Genetics at the School of Molecular and Biomedical Science at the University of Adelaide. His research focuses on the endosymbiotic coevolution of nuclear, mitochondrial and chloroplast genomes. He also investigates the genetic control of cotton fibre development with a view to crop improvement. Timmis has published over 80 papers, including four in the high-impact journal Nature. He is president of the Genetics Society of Australia and will present this year's MJD White address.

#### **Richard Harvey (Victor Chang Institute)**

Professor Richard Harvey is head of the Developmental Biology Program and Deputy Director at the Victor Chang Cardiac Research Institute. His research focuses on the Molecular mechanisms underlying formation, morphogenesis and function of the mammalian heart. He uses genomic and bioinformatic technologies to dissect genetic pathways in the developing mouse, which facilitates a better understanding of the molecular basis of cardiac disease. In March 2007, Richard was elected a Fellow of the Australian Academy of Sciences.

### John S Mattick (University of Queensland)

Professor John Mattick is Foundation Chair of the Institute of Molecular Biosciences at The University of Queensland. He was responsible for the development of one of the first recombinant DNA-based vaccines, and more recently has pioneered studies into the role of non-coding RNAs in the evolution and development of complex organisms. He is a member of the Queensland Biotechnology Advisory Council and is on the Scientific Advisory Boards of several institutes nationally and internationally. In 2001 he was appointed as an Officer in the Order of Australia.

### Scott O'Neill (University of Queensland)

Professor Scott O'Neill is Head of Integrative Biology at The University of Queensland. He was among the first researchers to apply molecular biological tools to understand the biology of the fascinating inherited bacterium *Wolbachia*, arguably the most common parasite in the world. Interest in *Wolbachia* is currently exploding, in part thanks to O'Neill's early work. His team recently finished sequencing the first *Wolbachia* genome, and is now using genomic tools to understand the molecular mechanisms by which it exerts its remarkable effects on host biology. This has led to the award of a \$10 m grant from the Gates foundation aimed at manipulating the ability of insect disease vectors to transmit pathogens to humans.

### Barry Brook (University of Adelaide)

Professor Brook is an international research leader in conservation biology and ecology. He has published two books and over 90 scientific papers on aspects of species preservation, extinction, conservation genetics, the effects of deforestation and climate change, and methods of sustainable wildlife management. In 2006, aged 31, he won the Australian Academy of Science Fenner Medal for distinguished research in biology and in 2007 was awarded the Edgeworth David Medal by the Royal Society of New South Wales. In March 2007 took up an appointment as the Foundation Sir Hubert Wilkins Chair of Climate Change and Director of the Research Institute for Climate Change and Sustainability at the University of Adelaide. The principal motivation for his research is to identify ways and means of reducing extinctions and mitigating the worst ravages of global change.

#### Marilyn Renfree (University of Melbourne)

Professor Marilyn Renfree's primary research interest is the developmental biology, reproductive physiology and endocrinology of mammals and she has worked on a wide range of species from mice to elephants but most of her research has been on marsupials, because of their ingenious alternative solutions to reproduction. Marsupials give birth to small underdeveloped young that undergo the majority of their development in the pouch, making it possible to study the control of normal organ growth throughout developmental stages otherwise inaccessible in utero. Her laboratory is known internationally for its innovative studies of these unique and charismatic Australian animals. Marilyn is a Laureate Professor of the University of Melbourne, an ARC Federation Fellow, the Ian Potter Chair of Zoology and Deputy Director of the ARC Centre of Excellence for Kangaroo Genomics.

#### **Bill Ballard (University of New South Wales)**

Professor John William Oman Ballard is an interdisciplinary scientist with skills in mitochondrial evolution, population genetics and bioenergetics. He has published 55 peer reviewed papers and been cited over 1,000 times. Ballard has received funding from NHMRC, NIH, NSF and private industry. Significantly, he was awarded an unsolicited Special NSF Creativity Extension Award in 2000. Ballard received his PhD from the University of Queensland in 1990 and was awarded a CSIRO postdoctoral fellowship in the same year. He was a Postdoctoral Fellow at the University of Chicago from 1991-94 and faculty at Field Museum in Chicago from 1995-2001. Ballard moved to the University of Iowa in 2001, as Professor and Foundation Director of the Carver Center of Comparative Genomics, and in 2006 accepted the position of Professor of Evolutionary Genetics in the School of Biotechnology and Bimolecular Science (BABS) at University of New South Wales.

# Symposia + topics

- Speciation and Phylogeography
- Adaptation
- Evolutionary Genetics
- Phylogeny, Biodiversity & Barcoding
- Developmental Genetics
- Epigenetics
- Population Genetics
- Pathogens, Parasites & Symbionts
- Environmental Microbes
- Plant Genetics
- Functional Genomics
- Comparative Genomics
- Bioinformatics
- Ancient and Forensic DNA
- RNAi and non-coding DNA
- Conservation Genetics
- Gene and QTL Mapping

# PROGRAM

# Tuesday 26th June, 2007

| P1                      | 09:00 -<br>09:45                     | Differentiation and development: The marsupial solution.  |  |  |  |  |  |
|-------------------------|--------------------------------------|---|--|--|--|--|--|
|                         | 09:45                                | Marilyn Renfree   |  |  |  |  |  |
| Plenary                 | 09:45 -                              | Genomes and Genetic Diversity in Primates and People  |  |  |  |  |  |
| Webster Lecture Theatre | 10:30                                | Richard A Gibbs   |  |  |  |  |  |
|                         |                                      |   |  |  |  |  |  |
| Γ1                      | 11:00 -                              | Identifying genes encoding symbiotic transporters in nitrogen-fixing soybean                                    |  |  |  |  |  |
|                         | 11:15                                | David Day   |  |  |  |  |  |
| Comparative             |                                      |   |  |  |  |  |  |
| Genomics 1              | 11:15 -<br>11:30                     | A lousy genome! The 110 Mb genome of the human body louse, Pediculus humanus                                    |  |  |  |  |  |
| Webster Lecture Theatre |                                      | Stephen Barker  |  |  |  |  |  |
|                         | 11:30 -<br>11:45                     | Evolution of sex chromosomes in snakes  |  |  |  |  |  |
|                         |                                      | Denis O'Meally  |  |  |  |  |  |
|                         | 11:45 -<br>12:00<br>12:00 -<br>12:15 | Sex microchromosomes in Australian dragon lizards   |  |  |  |  |  |
|                         |                                      | Tariq Ezaz  |  |  |  |  |  |
|                         |                                      | Genome organisation in platypus and chicken sperm   |  |  |  |  |  |
|                         | 12:15                                | Frank Grützner  |  |  |  |  |  |
|                         | 12:15 -<br>12:30                     | The origin of platypus venom molecules  |  |  |  |  |  |
|                         | 12:30                                | Camilla Whittington   |  |  |  |  |  |
|                         | 12:30 -<br>12:45                     | Characterization of divergent immune gene families in a marsupial and a<br>monotreme                            |  |  |  |  |  |
|                         |                                      | Emily Wong  |  |  |  |  |  |
|                         |                                      |   |  |  |  |  |  |
| 22                      | 14:00 -<br>14:45                     | The chicken genome: an evolutionary tool for the identification of functional elements in the vertebrate genome |  |  |  |  |  |
| Plenary                 |                                      | David W Burt  |  |  |  |  |  |
| Valatar Lasting Theoton | 14:45 -                              | Tools for comparative genomics  |  |  |  |  |  |
| Webster Lecture Theatre | 15:30                                | Frank Nicholas  |  |  |  |  |  |

# **Genetics Society of AustralAsia Meeting**

| Р3                            | 18:00 -<br>18:05 | Opening Address<br>Merlin Crossley  |
|-------------------------------|------------------|---|
| Catcheside<br>Prize           | 18:05 -<br>18:25 | An ENU screen for modifiers of epigenetic reprogramming reveals novel sex-<br>specific and parental effects                           |
| Webster Lecture<br>Theatre    |                  | Marnie Blewitt  |
| S1                            | 18:25 -<br>18:40 | Testing estimates of genetic population structure: levels of confidence in population genetics  |
| Student                       |                  | Clare E Holleley  |
| Presentations Webster Lecture | 18:40 -<br>18:55 | Devil Facial Tumour Disease: Lack of histocompatibility barriers explains the spread of a transmissible tumour                        |
| Theatre                       |                  | Hannah V Siddle   |
|                               | 18:55 -<br>19:10 | Characterisation of the beta-globin cluster and its flanking regions in the egg-laying monotreme, Ornithorhynchus anatinus (platypus) |
|                               |                  | Vidushi S Patel   |
|                               | 19:10 -<br>19:25 | Impact of Heavy-Metal Pollution on Sediment Microbial Community Diversity in an Urban Creek   |
|                               |                  | Carly P Tucker  |
|                               | 19:25 -<br>19:40 | Insight into the limits of the Japanese encephalitis virus through a better understanding of the biodiversity of its mosquito vector. |
|                               |                  | Stéphane Hemmerter  |

# Wednesday 27<sup>th</sup> June 2007

# Joint Genetics Society and Comparative Genomics Workshop

| P4                         | 09:00 | Genome evolution in Reptilia, the sister group of mammals |
|----------------------------|-------|---|
| Plenary                    |       | Scott Edwards   |
| ,                          | 09:45 | Real progress in Genome to Phenotype                      |
| Webster Lecture<br>Theatre |       | Win Hide  |

| W1      | 11:00 -          | Genomics and             | W2          | 11:00   | Unexpectedly ancient                         | W3               | 11:00   | Comparison of multiple        |
|---------|------------------|--------------------------|-------------|---------|--|------------------|---------|-------------------------------|
|         | 11:15            | metagenomics of          |             | - 11:15 | cryptic species and                          |                  | - 11:15 | sequence alignment            |
| C       |                  | marine and Antarctic     |             |         | global oceanic dispersal                     | B                |         | methods, with                 |
| 0       |                  | microorganisms           | Н           |         | in the smallest                              | I                |         | implications for surveys      |
| M       |                  | Torsten Thomas           | Y           |         | eukaryotes                                   | 0                |         | of proteomic data             |
| P       |                  |                          | L           |         | Jan Slapeta                                  | I                |         | Tanya Golubchik               |
| A       | 11:15 -          | Meta-analysis of         | 0           | 11:15   | Mitochondrial and                            | N<br>F           | 11:15   | Compositional                 |
| R       | 11:30            | metagenomes:             | G<br>E      | - 11:30 | ribosomal DNA spacer                         | Г<br>О           | 11:30   | Heterogeneity and             |
| A<br>T  |                  | metabolic                | E<br>O      | 11100   | evolution of the SW                          | R                |         | Model Misspecification        |
| I       |                  | fingerprints of          | G           |         | Pacific malaria vector                       | M                |         | in Phylogenetic Studies       |
| V       |                  | microbial and viral      | R           |         | A. farauti s.s. reveals a                    | A                |         | Kwok Wai Lau                  |
| Ē       |                  | communities from         | A           |         | complex demographic                          | T                |         |                               |
|         |                  | eight environments.      | P           |         | history and interesting                      | I                |         |                               |
| G       |                  | Elizabeth A Dinsdale     | H           |         | concerted evolution.                         | Ĉ                |         |                               |
| E       |                  |                          | Y           |         | Nigel W Beebe                                | S                |         |                               |
| N       | 11:45 -<br>12:00 | Evolution of             |             | 11:45   | Phylogeography of the                        |                  |         | CpG substitution rate         |
| 0       | 12.00            | segmented                |             | 12:00   | Amazonian Pencilfish                         | Clunie<br>s Ross | 12:00   | variation in mammalian        |
| M       |                  | mitochondrial            | HR<br>Carne |         | Nannostomus                                  | Lectur           |         | genomes                       |
| I       |                  | genomes in the lice of   | Lectur      |         | unifasciatus                                 | e                |         | Helen Lindsay                 |
| C       |                  | humans and other         | e           |         | (Lebiasinidae) Using                         | Theatr           |         |                               |
| S       |                  | primates                 | Theatr<br>e |         | Intron DNA Markers                           | c                |         |                               |
|         |                  | Renfu Shao               | ľ           | 10.00   | Mark J Sistrom                               |                  | 10.00   |                               |
| 2       | 12:00 -<br>12:15 | Evolution of             |             | 12:00   | Comparative                                  |                  | 12:00   | Gentrepid: A                  |
| Webster |                  | Cytochrome P450          |             | 12:15   | phylogeography of two                        |                  | 12:15   | Bioinformatics System         |
| Lecture |                  | Structures               |             |         | long-necked turtle                           |                  |         | For Candidate Disease         |
| Theatre |                  | Bagavananthem            |             |         | species in the Murray-<br>Darling Basin: How |                  |         | Gene                          |
|         |                  | Andavan Gowri<br>Shankar |             |         | molecular data can                           |                  |         | Merridee A Wouters            |
|         |                  | Snankar                  |             |         | answer ecological                            |                  |         |                               |
|         |                  |                          |             |         | questions                                    |                  |         |                               |
|         |                  |                          |             |         | Kate Hodges                                  |                  |         |                               |
|         | 12:15 -          | Know your enemy –        |             | 12:15   | Using ancient DNA to                         |                  | 12:15   | A grid of Oxford grids:       |
|         | 12:30            | Detailed analysis of     |             | -       | assess phylogeography                        |                  | -       | a comparative genomics        |
|         |                  | the the genome of the    |             | 12:30   | of European, Russian                         |                  | 12:30   | tool for Ensembl's            |
|         |                  | sheep blowfly,           |             |         | and North American                           |                  |         | automatically annotated       |
|         |                  | Lucilia cuprina          |             |         | brown bears (U. arctos).                     |                  |         | genomes                       |
|         |                  | Philip Batterham         |             |         | Sarah Bray                                   |                  |         | Jonathan M. Usmar             |
|         | 12:30 -          | Natural Killer           |             | 12:30   | From populations to                          |                  | 12:30   | Modelling the Omics           |
|         | 12:45            | Complex Genes In         |             | -       | species: the influence of                    |                  | -       | Network of                    |
|         |                  | Marsupials And           |             | 12:45   | ecological and                               |                  | 12:45   | Hepatocellular                |
|         |                  | Monotremes               |             |         | environmental factors                        |                  |         | <b>Carcinoma Using Visual</b> |
|         |                  | Claire Elizabeth         |             |         | on rainforest diversity                      |                  |         | Graph Mining                  |
|         |                  | Sanderson                |             |         | Maurizio Rossetto                            |                  |         | David Cho Yau Fung            |
|         | 12:45 -          | Class I MHC genes in     |             | 12:45   | Fine-scale phylo-                            |                  | 12:45   | Comparison of false           |
|         |                  | the tammar wallaby       |             | -       | geographic structure in                      |                  | -       | discovery rate                |
|         |                  | (M. eugenii) are         |             | 13:00   | an Amazonian flooded-                        |                  | 13:00   | controlling procedures        |
|         |                  | dispersed throughout     |             |         | forest ecosystem:                            |                  |         | for microarrays               |
|         |                  | the genome               |             |         | a multispecies                               |                  |         | Jie Song                      |
|         |                  | Hannah V Siddle          |             |         | comparative study                            |                  |         |                               |
|         |                  |                          |             |         | Luciano B Beheregaray                        |                  |         |                               |
|         |                  |                          |             |         |  |                  |         |                               |

# Wednesday 27<sup>th</sup> June 2007 continued

| W4                              |                     |   | W5  |                     | Molecular markers for   | W6   | 14:00               | Deletion of msh-2 in   |
|---------------------------------|---------------------|---|---|---------------------|---|--|---------------------|--|
| C<br>O<br>M<br>P<br>A           | 14:00<br>-<br>14:15 | The Kangaroo<br>Genome - Filling The<br>Phylogenetic Gap<br>Elizabeth Kuczek  | P<br>H<br>Y<br>L<br>O                     | -                   | understanding the<br>evolutionary and<br>population genetics of<br>the pest moth genus<br>Helicoverpa<br>Wee Tek Tay        | F<br>U<br>N<br>C<br>T                      | 14:15               | Neurospora: expected<br>phenotype with some<br>surprises<br>P Jane Yeadon  |
| R<br>A<br>T<br>I<br>V<br>E      |                     | Mapping genes on<br>tammar wallaby<br>chromosome 5<br>Janine E Deakin   | G<br>E<br>N<br>Y<br>HR<br>Carne<br>Lectur | 14:15<br>-<br>14:30 | medically important<br>Australian Chrysomya<br>(Diptera:<br>Calliphoridae)  | I<br>O<br>N<br>A<br>L<br>G                 | -                   | <b>Dominance in absentia –</b><br><b>phantom rec+ genes.</b><br><i>Frederick J Bowring</i>   |
| G<br>E<br>N<br>O<br>M<br>I<br>C | -                   | Cytogenetic and<br>inactivity map of the<br>tammar wallaby X<br>chromosome<br>Edda Koina  | Lectur<br>e<br>Theatr<br>e                | 14:30<br>-<br>14:45 | Leigh A Nelson<br>Evolution of Optical<br>Microstructures on the<br>Wings of Butterflies<br>Julia C Jones                   | E<br>N<br>O<br>M<br>I<br>C<br>S            | 14:30               | The role of a new class of<br>transcriptional activator<br>in the response to nutrient<br>limitation and<br>programmed cell death<br>Margaret E. Katz  |
| S<br>3<br>Webster<br>Lecture    | 14:45<br>-<br>15:00 | Opossum X Chromo-<br>some sequence reveals<br>steps in the evolution<br>of the human X And X<br>inactivation<br>Matthew J Wakefield |   | 14:45<br>-<br>15:00 | Genetic divisions of the<br>Collared peccary from<br>the Americas<br>Jaime Gongora  | 1<br>Clunies<br>Ross<br>Lecture<br>Theatre | -<br>15:00          | Molecular factors<br>involved in Thielaviopsis<br>basicola-plant<br>interactions<br>Lily Pereg-Gerk  |
| Theatre                         | -                   | The evolution of genes<br>regulating genomic<br>imprinting in<br>mammals.<br><i>Timothy A Hore</i>                                  |   | 15:00<br>-<br>15:15 | Screening For Porcine<br>Endogenous<br>Retrovirus Sequences<br>Among Species Of The<br>Family Suidae<br>Fabrícia Nascimento |  | 15:00<br>-<br>15:15 | Adaption in the Hawaiian<br>Picture-winged<br>Drosophila<br>Marla A Fisher   |
|                                 | 15:15<br>-<br>15:30 | Which came first:<br>Mammals or the X<br>and Y?<br>Paul D Waters  |   | -                   | Ancient DNA sheds<br>light on moa gigantism<br>and dwarfism in New<br>Zealand<br>Nic Rawlence                               |  | 15:15<br>-          | Investigating the target<br>sites of Phenylpyrazole<br>insecticides in Drosophila<br>melanogaster<br><i>Emily J Remnant</i>  |
|                                 | 15:30<br>-<br>15:45 | Use of Comparative<br>Genomics in<br>Identifying Disease<br>Genes in Dogs<br>Alan N Wilton  |   | 15:30<br>-          | Evolutionary<br>conservation of<br>microsatellites in<br>mammalian genomes<br>Emmanuel Buschiazzo                           |  | 15:30<br>-<br>15:45 | Generation and<br>characterisation of<br>resistance to<br>neonicotinoids in<br>Drosophila melanogaster<br>Trent Perry  |
|                                 | 15:45<br>-<br>16:00 | Building a virtual<br>genome using<br>comparative genomics<br>and limited genome<br>sequencing<br>Brian Paul Dalrymple              |   |                     |   |  | 15:45<br>-<br>16:00 | Challenging strategies in<br>development of cost<br>effective methods from<br>collection and storage of<br>blood samples to high-<br>throughput DNA<br>extraction<br>Mohammad Reza Shariflou |

| P5 Plenary                 | 16:30 | Phenomics: It is a team effort                          |
|----------------------------|-------|---|
| Webster Lecture<br>Theatre | 17:15 | Bill Ballard  |
|                            | 17:15 | The Trail to the Trial: A Cat Hair and Feline Forensics |
|                            | 18:00 | Marilyn Menotti-Raymond                                 |

| Thursday 28 <sup>th</sup> June 2007     |                  |  |  |  |  |  |
|---|------------------|--|--|--|--|--|
| Genetics Society of AustralAsia Meeting |                  |  |  |  |  |  |
| P6 Plenary                              | 09:00 -<br>09:45 | Creating complex antibiotic resistance gene regions; many resistance genes and many mechanisms |  |  |  |  |
| Webster Lecture<br>Theatre              |                  | Ruth M Hall  |  |  |  |  |
| Theate                                  | 09:45 -<br>10:30 | Wolbachia-mediated life shortening of Aedes aegypti to reduce dengue transmission              |  |  |  |  |
|   |                  | Scott O'Neill  |  |  |  |  |

| Th1<br>P<br>O                 | 11:00<br>-<br>11:15 | A Genetic Marker for<br>the Queensland Fruit<br>Fly in SIT programs<br>Jennifer Morrow   | Th2<br>E<br>V                        | 11:00<br>-<br>11:15 | What Drives<br>Evolution?<br>Rolf Beilharz  | Th3<br>F<br>U                             | 11:00<br>-<br>11:15 | Molecular evolution and<br>the transition to<br>viviparity<br>Bridget Frances Murphy   |
|-------------------------------|---------------------|--|--------------------------------------|---------------------|---|---|---------------------|--|
| P<br>U<br>L<br>A              | 11:15<br>-<br>11:30 | Molecular character-<br>isation of microsat<br>DNA and EPIC DNA<br>markers of the pest   | O<br>L<br>U<br>T                     | 11:15<br>-<br>11:30 | Two independent<br>duplications forming<br>the Cyp307a genes in<br>Drosophila.  | N<br>C<br>T<br>I                          | 11:15<br>-<br>11:30 | A potent antimicrobial<br>protein expressed in<br>wallaby milk<br>Benjamin G Cox   |
| T<br>I                        |                     | moth H. armigera<br>Wee Tek Tay  | I<br>O                               |                     | Tamar Sztal   | O<br>N                                    |                     | Benjamin G Cox   |
| O<br>N<br>G<br>E              | 11:30<br>-<br>11:45 | Racial origin of<br>commercial and feral<br>honey bees (Apis<br>mellifera) in WA.  | N<br>A<br>R<br>Y                     | 11:30<br>-<br>11:45 | Mechanisms of gene<br>transfer from the<br>chloroplast to the<br>nucleus  | A<br>L<br>G<br>E                          | 11:30<br>-<br>11:45 | Investigation of the<br>immune strategies for<br>survival in the neonatal<br>tammar (M. eugenii).  |
| N<br>E<br>T<br>I<br>C         | -<br>12:00          | Nadine C Chapman<br>Oceanic variability<br>and coastal<br>topography shape<br>local genetic structure                              | G<br>E<br>N<br>E<br>T                | 12:00               | Sven K. Delaney<br>Instability of<br>chloroplast-derived<br>DNA in the nuclear<br>genome of tobacco                               | N<br>O<br>M<br>I<br>C                     | 11:45<br>-<br>12:00 | Kerry Daly<br>A comparative genomics<br>aproach to identify<br>lactation candidate genes<br>Palaniappan Ramanathan                           |
| S<br>1                        |                     | in a long-dispersing<br>marine invertebrate<br>Sam Banks   | I<br>C<br>S                          |                     | Anna Sheppard   | S<br>2                                    |                     |  |
| Webster<br>Lecture<br>Theatre |                     | The genetic and<br>behavioural plasticity<br>of montane<br>populations   | HR<br>Carne<br>Lectur<br>e<br>Theatr | -                   | Sex linked genetic<br>influence on caste<br>determination in a<br>termite   | Clunie<br>s Ross<br>Lectur<br>e<br>Theatr |                     | Expression of myostatin<br>in cattle selected for high<br>and low muscling and in<br>cattle heterozygous for a<br>myostatin loss of function |
|                               |                     | Kate Umbers  | e                                    |                     | Nathan Lo   | e   |                     | myostatin loss of function<br>mutation<br>Grant Peter Parnell  |
|                               | 12:30               | Population structure<br>of the Giant<br>Australian Cuttlefish -<br>implications for the<br>world's largest<br>breeding aggregation |                                      | 12:30               | Did a lineages of<br>Pediculus (lice)<br>evolve on neander-<br>thals? Are the head<br>lice and body lice of<br>humans the same or |   |                     | A novel role for Xist RNA<br>in the formation of a<br>repressive nuclear<br>compartment into which<br>genes are recruited when<br>silenced.  |
|                               |                     | of cephalopods<br>Steve Donnellan  |                                      |                     | different species?<br>Natalie Leo   |   |                     | Julie Chaumeil   |
|                               | -<br>12:45          | Pop genetics and<br>breeding in the Giant<br>Gippsland earthworm,<br>Megascolides australis  |                                      |                     | Cane toad (Bufo<br>marinus) toxin<br>resistance in goannas<br>Beata Ujvari  |   | 12:30<br>-<br>12:45 | Moderate effects of<br>myostatin F94L on cattle<br>carcass traits<br>C D.K. Bottema  |
|                               | 12:45<br>-<br>13:00 | Neil Murray<br>Immunocontraception<br>of mammalian<br>wildlife: ecological &<br>immunogenetic issues<br>Desmond W Cooper           |                                      | -<br>13:00          | Reduced microsat<br>variation on a<br>marsupial Y<br>chromosome<br>Anna J MacDonald   |   |                     |  |
|                               |                     | Desmona w Cooper   |                                      |                     | Annu J MucDonula  |   |                     |  |

| Genetic structure of blue whales<br>(Balaenoptera musculus) in the<br>Southern Hemisphere | The cyst-forming nematode,<br>Globodera rostochiensis, has a<br>multipartite mitochondrial | Candidate gene identification by<br>combining QTL mapping with<br>bioinformatics      |  |  |
|---|--|---|--|--|
| Catherine Attard  | genome<br>Tracey Gibson  | Jennifer A Saleeba  |  |  |
| GEM DB: Griffith Ensembl<br>Mirror Database   | Differential evolution of classical  | Phylogeography, conservation<br>genetics and management of estuary                    |  |  |
| Kimberly N Begley   | and non-classical class I genes of the major histocompatibility                            | perch (Macquaria colonorum)<br>Kim Shaddick   |  |  |
| Function and Regulation of the<br>Drosophila melanogaster                                 | complex among suids and peccaries  | Inferring the Protein Domain  |  |  |
| <b>Cytochrome P450 gene Cyp12d1</b><br>Adrian Boey  | Jaime Gongora  | Boundary of Discontinuous Domain<br>Using DomainDiscovery and Inter-                  |  |  |
|   | Functional characterisation of   | <b>Residue Contact Interactions</b>   |  |  |
| A comparative genomics<br>approach to identifying   | Cytochrome P450 functions in<br>Drosophila melanogaster by                                 | Abdur Rahman Sikder   |  |  |
| endosymbiont loci associated with<br>early death in insects                               | genetic knockout and RNAi<br>Thomas W. Harrop  | Comparative Mapping And Analysis<br>Of Breakpoint Regions Between                     |  |  |
| Jeremy C Brownlie   | Ankyrin Domain Proteins:   | Ovine And Bovine Chromosomes<br>Using Radiation Hybrid Mapping                        |  |  |
| Investigating how a classic reproductive parasite might also                              | Abundant, Variable And Useful<br>In Understanding The Wolbachia-                           | Mini Singh<br>On Extending the Hardy-Weinberg   |  |  |
| act as a mutualist  | Insect Symbiosis<br>Iñaki Iturbe-Ormaetxe  |   |  |  |
| Jeremy C Brownlie   | Inaki liurbe-Ormaeixe  | Law<br>Alan Edmund Stark  |  |  |
| Understanding the nature of host adaptation for a common                                  | Phylogenetic characterisation of cycad-cyanobacteria symbiosis                             | Mating structure and inter-nest   |  |  |
| endosymbiont through<br>comparative genomics  | Marilena Meloni  | relatedness in Polistes wasps<br>Adam Stow  |  |  |
| Jeremy C Brownlie   | Microsatellites in the Yeast<br>Genome   | Expression Analysis of Cytochrome   |  |  |
| Estimating the propagule size of a  |  | P450s in Drosophila melanogaster  |  |  |
| recent outbreak<br>Emilie C Cameron   | Biogeographic Patterns Across  | Tamar Sztal   |  |  |
| Water colour and fish evolution:  | Three Tropical Butterfly Genera<br>(Lepidoptera) In The Indo-Pacific                       | Transposable elements in the tammar wallaby genome                                    |  |  |
| comparative phylogeography of   | Chris James Muller   | Katherine Thompson  |  |  |
| fishes from the major rivers of<br>Amazonia<br>Georgina M Cooke                           | Spawning color divergence<br>reflects local adaptation among                               | Mapping the X-linked angiotensin<br>receptor 2 gene and X-Y pairing in                |  |  |
| Georgina in Cooke   |  | cattle, sheep and goats   |  |  |
| Evolutionary and Conservation<br>Genetics of Wobbegong Sharks                             | Kristina M Ramstad   | Graham C Webb   |  |  |
| ( <b>Orectolobidae</b> )<br>Shannon Corrigan  | Love is in the Air: MHC & Mate<br>Choice in the Cunnningham's<br>Skink                     | Exploring the functional organization<br>of eukaryotic genomes using Gene<br>Ontology |  |  |
| A User Study on Two Gene  | Vincenzo Repaci  | Rohan BH Williams   |  |  |
| <b>Ontology Visualization Methods</b><br>David Cho Yau Fung                               | The Unusual Mitochondrial  | The influence of male development   |  |  |
| Classification of Expression  | Genome of the Potato-Cyst<br>Nematode, Globodera   | time on strength of Wolbachia-<br>induced CI expression in Drosophila                 |  |  |
| Profiles based on Visual Graph<br>Topology Analysis                                       | <b>rostochiensis</b><br>Angelique Helena Riepsamen   | melanogaster<br>Ryuichi Yamada  |  |  |
| David Cho Yau Fung  |  |   |  |  |

# Plenary Session

| P7 Plenary      | 16:30 - 17:15 | Using Ancient DNA to analyse assumptions used in evolutionary reconstruction |
|-----------------|---------------|--|
| Webster Lecture |               | Alan Cooper  |
| Theatre         | 17:15 - 18:00 | Evolutionary adaptation to climate change                                    |
|                 |               | Barry W. Brook   |

# Friday 29<sup>th</sup> June 2007

# **Genetics Scoiety of AustralAsia Meeting**

| P8 Plenary                 | 09:00 -          | Genetic pathway defects underlying congenital heart disease  |
|----------------------------|------------------|--|
|                            | 09:45            | Richard Harvey   |
| Webster Lecture<br>Theatre | 09:45 -<br>10:30 | The majority of the genomes of complex organisms encode regulatory RNAs that control differentiation and development |
|                            |                  | John S Mattick   |

|              | 11.0        |  |                   | 11.0        |   |                    | 11.0        |                         |
|--------------|-------------|--|-------------------|-------------|---|--------------------|-------------|-------------------------|
| F1           |             | <b>MEGMAR:</b> predicting                                      | F2                | 11:0        | Effects of an invasive                          | F3                 | 0 -         | Mapping the Gene for    |
|              | 11:1        | connectivity in the sea  |                   | 11:1        | toxic prey on the genetics                      |                    | 11:1        | Worker Sterility in the |
| P            | 5           | using codistributed  | C                 | 5           | of a naive native squam-                        | M                  | 5           | Honey Bee (Apis         |
| 0            |             | population data sets   | 0                 |             | ate predator: cane toads                        | A                  |             | mellifera)              |
| P            |             | Luciano B Beheregaray  | N                 |             | and goannas in the NT                           | P<br>P             |             | Peter Robert Oxley      |
| U            |             |  | S                 |             | Thomas Madsen                                   | I P                |             |                         |
| L            | 11:1        | Genetic Diversity in   | E                 | 11:1<br>5 - | Genetic variation in                            | N                  | 11:1<br>5 - | QTL mapping for heavy   |
| A<br>T       | 5 -<br>11:3 | Laticaudid sea kraits :  | R<br>V            | 11:3        | koalas on French and                            | G                  | 11:3        | metal tolerance in      |
| I            | 0           | Is philopatry resulting  |                   | 0           | Kangaroo Island and the                         |                    | 0           | chironomids             |
| 0            |             | in genetic divergence?   | A<br>T            |             | likely effect of                                | Clunies            |             | Cintia M. Martinez-     |
| N            |             | Amanda Lane  | I                 |             | contraception protocols                         | Ross               |             | Garduno                 |
| 1            |             |  | Ó                 |             | on its retention                                | Lecture<br>Theatre |             |                         |
| G            |             |  | N                 |             | Romane Cristescu                                | Incure             |             |                         |
| E            | 11:3        | An Update on   |                   | 11:3        | Reduced MHC class II                            |                    | 11:3        | Towards a               |
| N            | 0 -<br>11:4 | Inbreeding in  | G                 | 0 -<br>11:4 | diversity in island                             |                    | 0 -<br>11:4 | Comprehensive Genetic-  |
| E            | 5           | Australian   | E                 | 5           | compared to mainland                            |                    | 5           | Linkage Map for the     |
| T            |             | Thoroughbred   | N                 |             | populations of a                                |                    |             | Saltwater Crocodile     |
| Î            |             | Racehorses   | E                 |             | threatened rock-wallaby                         |                    |             | (Crocodylus porosus)    |
| Ċ            |             | Kao Castle   | T                 |             | (P. lateralis lateralis)                        |                    |             | Lee Miles               |
| S            |             |  | Î                 |             | Robert A Mason                                  |                    |             |                         |
| Ŭ            | 11:4        | Occurrence of  | Ċ                 | 11:4        | Genetic variation in                            |                    | 11:4        | Map Integration And     |
| 2            | 5 -         | Hybridication between  | S                 | 5 -         | translocated northern                           |                    | 5 -         | Genome Comparison       |
|              | 12:0        | Sympatric Populations  |                   | 12:0        | quoll (Dasyurus                                 |                    | 12:0        | Involving The Tammar    |
| Webste       | Ū           | of Grey Kangaroo   | 1                 |             | hallucatus) island                              |                    | Ŭ           | Wallaby                 |
| r<br>Lecture |             | Linda E Neaves   |                   |             | populations.                                    |                    |             | Chenwei Wang            |
| Theatre      |             |  | HR                |             | Maria J Cardoso                                 |                    |             | 0                       |
|              | 12:0        | A population genetic   | Carne             | 12:0        | Bigger is not better: male                      |                    | 12:0        | Tammar Wallaby          |
|              | 0 -         | study of invasive fish   | Theatre           | 0 -         | reproductive success in a                       |                    | 0 -         | Constic Possures        |
|              | 12:1        | species, the common  |                   | 12:1        | captive breeding                                |                    | 12:1<br>5   | Des Cooper              |
|              | 5           | carp (C. carpio), in the                                       |                   | 5           | program for the greater                         |                    | 5           |                         |
|              |             | Murray-Darling   |                   |             | bilby (Macrotis lagotis)                        |                    |             |                         |
|              |             | Gwilym David Haynes  |                   |             | Emily J Miller                                  |                    |             |                         |
|              |             | Using linkage  |                   |             | An evaluation of                                |                    |             | A Comaprative Study Of  |
|              | 12:1        | disequilibrium to  |                   | 12:1        | estimating abundance                            |                    | 12:1        | The Abc-Family          |
|              | 5 -         | estimate effective   |                   | 5 -         | with faecal and hair DNA                        |                    |             | Transporter, ABCG2, As  |
|              |             | separation times for   |                   |             | for the endangered                              |                    | 0           | A Lactation Associated  |
|              |             | different worldwide  |                   |             | species the Spotted-tailed                      |                    |             | Candidate Gene          |
|              |             | human populations.   |                   |             | Quoll (D. maculatus)                            |                    |             | Peter Williamson        |
|              |             | John Sved  |                   |             | Monica P Ruibal                                 |                    |             | r erer withumson        |
|              | 12.3        | The Y chromosome as  |                   | 12.3        | An investigation of MHC                         |                    | 12.3        | A comparative and       |
|              |             |  |                   |             |   |                    | 0 -         | integrative genomics    |
|              | 12.4        | a tool in populations  |                   | 12:4        | diversity in koalas<br>(Phascolarctos cinereus) |                    | 12:4        | approach for            |
|              | 5           | genetics and disease   |                   | 5           |   |                    |             | identification of dairy |
|              |             | association studies  |                   |             | Sarah Elizabeth Jobbins                         |                    |             | QTL candidate genes     |
|              |             | Joanne Maree Lind  |                   |             |   |                    |             |                         |
|              | -           |  |                   | 10.4        |   |                    |             | Arun Jayappa            |
|              | 12.4        | Range Expansions,  |                   | 12:4        | Devil Facial Tumour                             |                    | 12:4        | Homozygosity mapping    |
|              | 5 -         | Rare Mutations And   |                   | 13:0        | Disease: cytogenetic clues                      |                    | 5 -         | of an recessive Rosai-  |
|              | 13:0        | Novel Environments:  |                   | 0           | to transmission and                             |                    |             | Dorfman-like condition  |
|              | 0           | <b>Evolution In Progress?</b>                                  |                   |             | development                                     |                    |             | in families using high- |
|              |             | Lee Ann Rollins  |                   |             | Hannah Sarah Bender                             |                    |             | density SNP genotyping. |
|              |             |  |                   |             |   |                    |             | Simon T Cliffe          |
| 10000        |             | a name a subsection of the subsection of the subsection of the | on all the second |             |   |                    |             |                         |

# Friday 29<sup>th</sup> June 2007 continued

| F4                                 | 14:00<br>14:15                   | Dramatic Variations in Microbial<br>Communities among Coral Reef   | F5   |  | Improving the mass-rearing of<br>Queensland Fruit Fly                           |  |
|------------------------------------|----------------------------------|--|--|--|---|--|
| C<br>O                             |                                  | Ecosystems<br>Elizabeth A Dinsdale   | A<br>D<br>A<br>P<br>T<br>A<br>T<br>I<br>O<br>N |  | Stuart Gilchrist  |  |
| N<br>S<br>E<br>R                   | 14:15<br>14:30                   | Pollen dispersal between planted<br>populations and remnant native<br>populations in a fragmented agricultural           |  | 14:15 -<br>14:30   | polymorphisms under thermal selection in Drosophila                             |  |
| V                                  |                                  | landscape<br>Margaret Byrne  |  |  | Ary Hoffmann  |  |
| A<br>T<br>I                        | 14:30<br>14:45                   | Plant Mating Systems And Assessing<br>Population Persistence In Fragmented   |  | 14:30 -<br>14:45   | An adaptive run at an insecticide resistance locus.                             |  |
| 0                                  |                                  | Landscapes<br>David Coates   | HR Carne                                       |  | Charles Robin   |  |
| N<br>G<br>N<br>E<br>T              | 14:45<br>15:00                   |  | Lecture<br>Theatre                             | 14:45 -<br>15:00   | Enigma of the yellow mutation of<br>the cotton bollworm Helicoverpa<br>armigera |  |
|                                    |                                  | Kioumars Ghamkhar  |  |  | Adam Williams   |  |
|                                    | 15:15<br>15:15<br>15:30<br>15:30 | Phylogeography of an endangered freshwater fish, Macquarie perch   |  | 15:00 -<br>15:15<br>15:15 -<br>15:30<br>15:30 -<br>15:45 | Wolbachia infection and insect activity   |  |
| I<br>C<br>S                        |                                  | (Macquaria australasica) in Eastern<br>Australia.  |  |  | Elizabeth McGraw  |  |
| 5                                  |                                  | Leanne K Faulks  |  |  |   |  |
| 2<br>Webster<br>Lecture<br>Theatre |                                  | Genetic diversity in Murray cod<br>(Maccullochella peelii peelii) across the<br>Murray-Darling Basin.                    |  |  | LDH-B and temperature<br>tolerance in coral trout<br>(Plectropomus leopardus)   |  |
|                                    |                                  | Meaghan L Rourke   |  |  | Emmanuelle Botte  |  |
|                                    |                                  | Grey nurse shark genetics: what do we know and what do we need to know?  |  |  | Amino Acid Site-Specific Fitness<br>and Epistasis in HIV-1                      |  |
|                                    |                                  | Heidi Ahonen   |  |  | Jack da Silva   |  |
|                                    | 15:45<br>16:00                   | Marked genetic differentiation in short-<br>beaked common dolphins subject to<br>fisheries impacts in southern Australia |  |  |   |  |
|                                    |                                  | Kerstin Bilgmann   |  |  |   |  |

| P9 Plenary              | 16:30 - 17:15 | Endosymbiotic evolution. |
|-------------------------|---------------|--------------------------|
| Webster Lecture Theatre |               | Jeremy N. Timmis         |

# **Social Events**

## **Public Forum**

Wednesday 27th June 2007 6pm for 6:30pm start, Halstrom Theatrette, The Australian Museum 45 mins panel discussion, and 15 minutes for audience questions. Followed by drinks until 9pm. Numbers are strictly limited to 140

# **Conference mixer**

Tuesday 26th June 3.30 to 5.45 pm to be followed by Conference opening at 6pm. The Grandstand Bar & Function Centre, University of Sydney Light snacks and drinks to be provided. Cost included in Registration Extra ticekts \$28 per person.

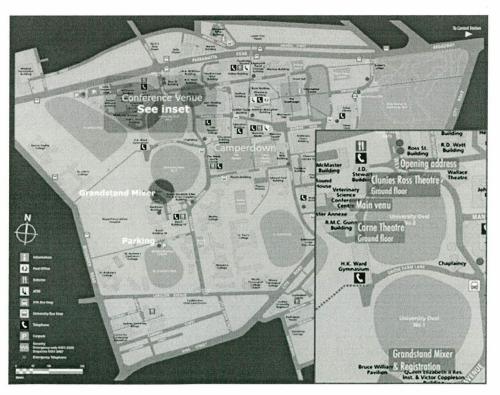
## **Student drinks**

Wednesday 27th June The Roxbury Hotel, 182 St John's Road, Glebe 6:30pm-8:30pm

The Student Drinks will be held at The Roxbury Hotel, which is situated a ten minute stroll from the conference venue. Come and join us for a relaxing evening of drinks and nibbles in the Mezzanine lounge at a Sydney Uni 'local'. This is a great opportunity to have a game of pool and a chat with other students and the GSA conference plenary speakers. Tickets \$20 per person.

# **Conference Dinner**

Thursday 28th June - 7pm to 11.30pm at Australian Museum, 6 College Street, Sydney. Dinner will be held in the Foyer. Live music will be provided by The Major Groove. Sponsored by the Australian Museum Cost: \$100 per person



# MAP

# **Public Forum - DNA and Crime**

# What?

# **DNA and Crime** at the Australian Museum

As part of the annual Genetics Society of Australasia conference, the Australian Musuem is hosting a panel discussion on the use of DNA to solve crimes. Believe it or not Australian Museum staff are frequently asked to assist with identifying animals that have been involved in crimes of varying description. We have several animal DNA experts on our panel as well as a human forensic expert. In a discussion led by ABCTV Catalyst's Paul Willis they will be talking about various cases they have been involved in.

# Who?

### should attend...

Want to know more? Then you should attend! Any one who is interested in using DNA to solve crime is invited. The event is free to members of the public but numbers are strictly limited.

# When?

### 6pm for 6:30pm start, 27th June 2007

Halstrom Theatrette, The Australian Museum 45 mins panel discussion, followed by 15 mins questions. Followed by drinks until 9pm. Numbers are strictly limited to 140

**Contact** Australian Museum Science Communications Unit (02) 9320 6389 or scicom@austmus.gov.au

# **Mediator**

Dr Paul Willis ABCTV Science Journalist

# Panel Members Dr Marylin Menotti-Raymond

Laboratory of Genomic Diversity, National Cancer Institute Frederick, Maryland

Dr. Menotti-Raymond is a staff scientist at the US National Cancer Institute-Frederick, MD. Her research has focused on the generation of genetic maps in the domestic cat in order to characterize genes associated with hereditary disease and related biological interest.

Dr. Raymond's group has recently mapped and characterized a mutation causative of late onset retinal atrophy in the Abyssinian cat, a model of human retinitis pigmentosa, and identified the causative mutation in a unique gene (LIX1) for spinal muscular atrophy. They have mapped and characterized mutations associated with coat color (chocolate, cinnamon, Burmese, Siamese) and the dilution of coat color (dilute) in the cat.

Additionally, Dr. Raymond has been involved with application of genetic markers in the cat for forensic analysis, which led to the first introduction of an animal DNA fingerprint into court and the development of an STR genotyping system for genetic individualization of cat specimens.

# Dr Roland van Oorschot

Forensic Services Department, Victoria Police

After a Agriculture degree in the Netherlands, a PhD in Australia on marsupial genetics, two years at the Southwest Foundation for Biomedical research in San Antonio Texas on gene mapping, and one year at the Centre for Animal Biotechnology in Melbourne on genetics of disease resistance in sheep, Dr van Ooschot started working, in 1992, at the Forensic Services Department of Victoria Police where he is currently the Manger of Research & Development and Quality Management of the Biology Division.

Dr van Ooschot has over 60 publications in scientific journals and books, Including an article in Nature in 1997, regarding the ability to retrieve DNA from touched objects, which has revolutionised forensic investigations and assisted in solving many thousands of cases world wide. His current interests are in the areas of: 'collection and typing of trace DNA samples' and 'getting more useful information from available DNA to assist criminal investigations'.

### **Dr Rebecca Johnson**

### DNA Laboratory Manager, Australian Museum

Dr Johnson is the manager of the Australian Museum's DNA Laboratory. Australian Museum staff are frequently asked to assist in identifying animals from their specialty area. These animals may have been involved in wildlife trafficking (live or dead), and/or in the trade of animal parts.

The work of the DNA laboratory is most useful when the animal parts no longer resemble the animal from which they came. Or if the animal is very underdeveloped and is difficult to identify as it doesn't have the characteristics of an adult specimens. DNA identification techniques have been very successfully usually used in these types of identifications. An example of projects the Australian Museum DNA laboratory has been involved with include: smuggled bird eggs, seized 'sea-horse' powder, identification of tuna and whale species from meat, identification of seized gall bladders, and identification of bird blood from a crime scene. Some of these crimes have resulted in \$10,000 fines or even time in jail.

# **Professor Stephen Donnellan**

### Evolutionary Biology Unit, South Australian Museum

Steve is a Principal Researcher at the South Australian Museum, Adelaide, Convenor of the South Australian Regional Facility for Molecular Evolution and Ecology at the University of Adelaide and Deputy Director of the Australian Centre for Evolutionary Biology & Biodiversity (ACEBB), University of Adelaide

He has more than 80 peer-reviewed scientific publications in the fields of evolutionary biology, molecular evolution, molecular ecology and systematics. His research has resulted in the discovery of more than 30 species of vertebrates, with a notable recent find of the third species of taipan, the most venomous group of snakes.

As a consequence of a research focus on biodiversity discovery and understanding the evolutionary relationships of the Australasian fauna, his research group has had a number of 'adventures' in wildlife forensics.

# **Colin Oxford**

Investigations Manager, Australian Customs Service

Colin is a Chief Investigator with the Australian Customs Service and leads a team which investigates, amongst other things, wildlife trafficking. He holds a Diploma of Government in Investigations and is a graduate of the Management of Serious Crime Course. He has 26 years experience with Customs with the last 13 spent in the Investigations Branch.

He has been involved in numerous investigations, which have led to the arrest of couriers attempting to smuggle bird eggs, venomous snakes and reptiles into and out of Australia as well as other exotic specimens used in traditional medicines. As a result he has experience in the DNA/Genetics requirements in Customs investigations.

On occasions bird smugglers will attempt to break eggs concealed on their bodies in the belief that this will render identification impossible. Of course they are wrong. Also, with worldwide concerns stemming from avian influenza many smuggled bird eggs have to be euthanased for quarantine reasons. As a result the most reliable method of identification of birds and other species is by way of DNA analysis. With many animals listed on the Appendices to the Convention on International Trade in Endangered Species (CITES) formal identification is also vital in preparing impact statements during criminal trials.

#### **Detective Inspector Russ Oxford**

### Homicide, NSW Police

Russell has spent 27 years in the NSW Police force and has extensive experience in the investigation of major crime, particularly homicide investigation.

# Plenary Abstracts (in alphabetical order)

WEDNSESAY 16:30 – 17:15 [P5 PLENARY] Phenomics: It is a team effort Bill Ballard<sup>1,\*</sup> <sup>1</sup>University of New South Wales Keywords: Functional Genomics Comparative Genomics

The suffix -omics generally refers to the study of a complete group or system of biomolecules. Genomics is the study of an organism's genome, proteomics is the study of an organism's entire complement of proteins and phenomics is the name given to the science which attempts to integrate the wealth of information into a holistic picture of the complete organism - its phenotype. Phenomics is the ambitious undertaking of integrative sciences in the forthcoming decade. At a clinical level, identification of the molecular bases of diseases and developmental disorders can have ramifications for diagnosis and intervention. Linking genotype to phenotype is also essential if we are to understand how natural genetic variation contributes to morphological trait variability in natural populations. Realizing these goals requires a diverse team of researches. I argue here, the pendulum of knowledge has swung towards a greater understanding of the genotype but has left the organismal phenotype at the far end of the arc.

THURSDAY 17:15 - 18:00 [P7 PLENARY]

**Evolutionary adaptation to climate change** Barry W. Brook<sup>1,\*</sup> <sup>1</sup>The University of Adelaide **Keywords:** Adaptation Conservation Genetics

Climate change – most particularly global heating – is one of the great environmental challenges facing society and natural systems in the 21st century. A strong emphasis in ecological climate change research, to date, has been on monitoring and interpreting the response of species and ecosystems to long-past and recent climate change. But what does the future hold? Recent attempts at projecting biodiversity scenarios over the coming centuries, at the broad scale, have concentrated on extrapolating bioclimatic envelopes and concomitant changes in habitat distributions. Yet a shifting geographical distribution is clearly only one mode of biological adaptation. I will review recent research on the evolutionary response of biota to climate change. I will particularly focus on the limits to adaptation of threatened and genetically impoverished species in the face of rapid global change.

### TUESDAY 14:00 - 14:45 [P2 PLENARY]

The chicken genome: an evolutionary tool for the identification of functional elements in the vertebrate genome David W Burt<sup>1,\*</sup>

<sup>1</sup>Roslin Institute **Keywords:** Comparative Genomics Functional Genomics

The chicken has long been an important model organism for developmental and evolutionary biology, as well as a major source of protein with billions of birds used in meat and egg production each year. Chicken genomics has been transformed in recent years, with the characterisation of large EST collections and most recently with the assembly of the chicken genome sequence. As the first bird genome to be sequenced it is a model for the remaining 9,600 species thought to exist today. Many of the features of avian biology and organisation of the chicken genome make it an ideal model organism for phylogenetics and embryology, along with applications in agriculture and medicine. The availability of new tools such as wholegenome gene expression arrays and SNP panels, coupled with information resources on the genes and proteins are likely to enhance this position.

THURSDAY 16:30 - 17:15 [P7 PLENARY] Using Ancient DNA to analyse assumptions used in evolutionary reconstruction Alan Cooper<sup>1,\*</sup> <sup>1</sup>University of Adelaide Keywords: Ancient and Forensic DNA Ancient and Forensic DNA Ancient DNA provides a unique means to observe evolution occurring in real time, and in general, has revealed evidence of a very dynamic and complex process that is seldom parsimonious. In other words, the detailed study of the current distribution and diversity of species and populations may reveal little about past evolutionary events. Ancient DNA has also identified many situations where the morphological interpretation of fossils does not match genetic data, and suggests that a range of new analyses into the epigenetic and QTL-basis of fossil morphological variation is required. Radiocarbondated bones of ancient animal populations have also provided a unique new means to calibrate rates of genetic change over short geological time periods, permitting the study of molecular clocks. This data has revealed considerable problems in the use of genetic sequences to date events in the recent past, and indicates that molecular clock studies may be inappropriate for small genetic distances. This includes important questions in human evolution, domestication, and conservation biology. The difficulties of dealing with damaged ancient DNA templates has also prompted the development of a range of new molecular methods suitable for the detection and accurate characterisation low copy number templates. A key recent innovation has been SPEX (Single Primer Extension) which provides a simple means to precisely examine the number and nature of starting templates in a PCR. This is likely to have a number of genetic and medical applications.

### WEDNESDAY 09:00 - 09:45 [P4 PLENARY] **Genome evolution in Reptilia, the sister group of mammals** Scott Edwards<sup>1,\*</sup> <sup>1</sup>Harvard University

Keywords: Comparative Genomics Evolutionary Genetics

With the draft chicken genome in hand and the draft genomes of Zebra Finch (Taeniopygia guttata) and the Greene Anole lizard (Anolis carolinensis) soon to be released, we are in a position to evaluate the broad differences in the dynamics of genome evolution between mammals on the one hand, and birds and non-avian reptiles on the other. In addition, we have been conducting large-scale end-sequencing of BAC libraries from a tuatara (Sphenodon punctatus), painted turtle (Chrysemys picta), Garter Snake (Thamnophis sirtalis), emu (Dromaius novaehollandiae) and American Alligator (Alligator mississipiensis) in order to add to the DNA sequence resources available for such study. This work suggests that non-avian reptiles possess a rich diversity of chicken-repeat 1 (CR1) family LINE elements and suggests that the ancestral amniote genome was also rich in these repeats, with drastic reductions occurring in the ancestor of extant birds. Using statistical analysis of the frequencies of 8mer oligonucleotides such as simplesequence repeats, we find that rates of interspecific oligonucleotidefrequency change in Reptilia are an order of magnitude slower than in mammals. We have begun to annotate extended regions of the major histocompatibility complex (MHC) in Anolis and Zebra Finch, as well as novel BAC sequences from alligator, turtle and emu that have been mapped to both sex chromosomes and autosomes using FISH and in silico. Alignments of these regions to available draft genomes suggest that, despite abundant small-scale intrachromosomal rearrangements, reptile genomies display a conservative mode of evolution commensurate with their slow rates of morphological change compared to mammals.

### TUESDAY 09:45 - 10:30 [P1 PLENARY] Genomes and Genetic Diversity in Primates and People Richard A Gibbs<sup>1,\*</sup> <sup>1</sup>Baylor College of Medicine Keywords: Comparative Genomics Comparative Genomics

Increased sequencing capacity, new technologies and the growth of genome data sets has begun to close the two decade old gap between 'genomics' and genetics. Illustrations of this convergence include the sequencing and analysis of the rhesus macaque genome, accompanied by analysis of SNPs in Indian and Chinese populations that reveal substantial bottlenecks in the former group; the sequencing of the bovine genome and development of breed specific markers; and most importantly the deep analysis of human variation in multiple individuals. The latter has led to the recent sequencing of a single human with next-generation sequencing technologies, pressing the challenge of how to interpret comprehensive data from a single person. The speed of increase of the power of the underlying nucleic acids technologies and the maturation of genome data sets bodes well for the prospect of a real impact of genomics on human health.

#### THURSDAY 09:00 - 09:45 [P6 PLENARY]

# Creating complex antibiotic resistance gene regions; many resistance genes and many mechanisms

Ruth M Hall<sup>1,</sup>

<sup>1</sup>School of Molecular and Microbial Biosciences, The University of Sydney, Sydney **Keywords:** Environmental Microbes Comparative Genomics

Both bacterial pathogens and commensal bacteria of humans and animals are often resistant to more than one antibiotic. The resistance genes are commonly carried together on plasmids or integrating elements that can move from one bacterium to another, where they are stably maintained. The resistance genes tend to cluster together forming resistance islands and several distinct types of translocational mechanism are involved in bringing different resistance genes together to create these complex clusters. The mechanisms used include transposition of transposons carrying resistance genes, site-specific integration of gene cassettes into integrons and a novel rolling circle based mechanism that picks up DNA adjacent to a novel type of small mobile element known as CR elements. There are now well over 100 different known gene cassettes that carry genes conferring resistance to a variety of antibiotics that are used in treatment of human disease or in animal husbandry, and the integron/gene cassette system can combine them together in many different ways. Members of the CR element family are now often found associated with a class 1 integron bringing in further resistance genes. Composite transposons created when two insertion sequences flank a resistance gene add to the mix. The evolution of resistance islands provides insight into the ways in which bacterial genomes evolve.

#### FRIDAY 09:00 - 09:45 [P8 PLENARY]

# Genetic pathway defects underlying congenital heart disease

Richard Harvey<sup>1,2,\*</sup> Owen WJ Prall<sup>1</sup>, Mary K Menon<sup>3</sup>, Mark J Solloway<sup>1</sup>, Stéphane Zaffran<sup>4</sup>, Fanny Bajolle<sup>4</sup>, Yusuke Watanabe<sup>4</sup>, Christine Biben<sup>1</sup>, Jim J McBride<sup>5</sup>, Bronwyn R Robertson<sup>2</sup>, Hervé Chaulet<sup>1</sup>, Fiona A Stennard<sup>1</sup>, Natalie Wise<sup>1</sup>, Hidetaka Shiratori<sup>6</sup>, Hiroshi Hamada<sup>6</sup>.,Brian L Black<sup>7</sup>,Yumiko Saga<sup>8</sup>, Elizabeth J Robertson<sup>9</sup>, Margaret E Buckingham<sup>4</sup>.

<sup>1</sup>Victor Chang Cardiac Research Institute, <sup>2</sup>University of New South Wales, <sup>3</sup>Victor Chang Cardiac Research Institute, <sup>4</sup>Pasteur Institute, <sup>5</sup>Garvan Institute of Medical Research, <sup>6</sup>Osaka University, <sup>7</sup>University of California, <sup>8</sup>National Institute of Genetics, Japan, <sup>9</sup>University of Oxford **Keywords:** Functional Genomics Developmental Genetics

Congenital heart disease (CHD) is the most common cause of non-infectious death in the first year of life. While we are beginning to assemble list of genes mutated in CHD, the genetic pathways underlying the heart development and affected in CHD are understood only in rudimentary detail. Nkx2-5 is a homeodomain transcription factor that sits high in the cardiac developmental hierarchy and physically associates with numerous other cardiac transcription factors and cofactors. It acts as a positive factor to support the cardiac transcriptional hierarchy, and to regulate chamber specification and growth, and conduction system development. Using transcript profiling, we show that the earliest function for Nkx2-5 is as a negative regulator of cardiac development and that disruption of this Nkx2-5-dependent negative feedback loop is causative in congenital heart disease (CHD). In Nkx2-5 null mice, cardiac induction and progenitor genes including Bmp2 are up-regulated in the heart progenitor fields and forming heart tube, leading initially to progenitor cell over-specification but subsequently a collapse of second heart field (SHF) proliferation and outflow tract (OFT) deployment. These events are evident in attenuated form in a hypomorphic Nkx2-5 model, which survives until birth showing compromised cardiac output and recapitulating NKX2-5-associated CHD. Elevated Bmp2/Smad1 signalling is a principal pathological mechanism in Nkx2-5 mutants - genetic deletion of Smad1 leads to SHF over-proliferation and deployment, and in null and hypomorphic Nkx2-5 mutants to rescued OFT development. Our findings show that the Nkx2-5/Bmp2/Smad1 pathway balances cardiac progenitor cell specification and proliferation, and forms a molecular switch ensuring separation of progenitor and differentiated states. The feedback loop is a potentially prevalent molecular target in CHD involving defects of the outflow tract.

#### WEDNESDAY 09:45 - 10:30 [P4 PLENARY]

#### Real progress in Genome to Phenotype

Win Hide<sup>F,\*</sup> Chris Maher<sup>1</sup> Adele Kruger<sup>1</sup> Vlad Bajic<sup>1</sup> Oliver Hofmann<sup>1</sup> <sup>1</sup>South African National Bioinformatics Institute, University of Western Cape, South Africa **Keywords:** Bioinformatics Functional Genomics Gene and QTL Mapping

Several exciting new studies have recently been published, as a result of genome-wide interrogation over tens of thousands of unrelated individuals. An example, that compares association scans in 17 000 healthy and afflicted individuals by the Welcome Trust Case Control Consortium, provides us with

new insight into human variation and disease. The scale of these studies is necessitated by the degree to which statistical power is required to derive meaningful results. Even upon resolution of genomic loci of interest to disease hunters, regions yield SNPs that may alter coding regions of genes; others lie in non-coding regions, and others in gene deserts, which contain no known genes. Clearly, our understanding of biological function can contribute a great deal to a reduction in the search space. In order to assess the value of candidate loci, an approach that resolves the transcript expression at that position, in the context of anatomy, time, species, cell type, and pathology, can be of great value. Candidate gene prioritization studies have begun to utilise these data, but with limited reliability. Given the degree to which gene expression data is available, a major contribution to its value is a better understanding of regulation and transcript isoform variation. We have embarked upon a process of mapping of regulation and isoform potential to gene expression and disease. The link between gene expression and phenotype is now beginning to yield real relationships. The extent to which an understanding of regulatory potential can yield disease relationships will be explored, using cancer stem cells and the macrophage as examples.

FRIDAY 09:45 - 10:30 [P8 PLENARY]

# The majority of the genomes of complex organisms encode regulatory RNAs that control differentiation and development

John S Mattick<sup>1,\*</sup> Larry J Croft<sup>1</sup> Marcel E Dinger<sup>1</sup> Michael Pheasant<sup>1</sup> Igor V Makunin<sup>1</sup> Marjan Askarian Amiri<sup>1</sup> Tim R Mercer<sup>1</sup>, Ken C Pang<sup>1,2</sup> Cas Simons<sup>1</sup> Ryan J Taft<sup>1</sup> <sup>1</sup>Institute for Molecular Bioscience, University of Queensland <sup>2</sup>Ludwig Institute for Cancer Research, Heidelberg **Keywords:** RNAi and non-coding DNA Functional Genomics

It appears that the genetic programming of the higher organisms has been fundamentally misunderstood for the past 50 years, because of the presumption - largely true in prokaryotes, but not in complex eukaryotes - that most genetic information is expressed as and transacted by proteins. The majority of the mammalian genome is in fact transcribed, apparently in a developmentally regulated manner, and most complex genetic phenomena in the higher organisms are RNA-directed. Evidence will be presented (i) that there are thousands of non-protein-coding transcripts in mouse that are dynamically expressed during differentiation and development, including in embryonal stem cell, muscle, genital ridge, macrophage, T-cell and neuronal cell differentiation, among others, many of which show precise expression patterns and subcellular localization in different regions of the brain; (ii) that there are millions of small RNAs expressed from the mouse and human genomes, the latter showing a massive expansion in the human brain; and (iii) that much, if not most, of the mammalian genome is not evolving neutrally, but comprises different types of sequences evolving at different rates under different selection pressures and different structure-function constraints. These observations suggest that the majority of the human genome and those of other complex organisms is devoted to a hidden RNA regulatory system that directs the trajectories of differentiation and development by controlling chromatin architecture and epigenetic memory, transcription, splicing, RNA modification and editing, mRNA translation and RNA stability.

WEDNESDAY 17:15 - 18:00 [P5 PLENARY]

**The Trail to the Trial: A Cat Hair and Feline Forensics** Marilyn Menotti-Raymond<sup>1,\*</sup> <sup>1</sup>National Cancer Institute-Frederick

Keywords: Ancient and Forensic DNA Population Genetics

DNA fingerprinting as a forensic tool has seen wide application in human genetic individual identification for well over a decade. Simple tandem repeat (STR) loci have become the polymorphic markers of choice for DNA typing applications. Hair can be a valuable source of forensic material at crime scenes and PCR genotyping of genomic and mitochondrial DNA extracted from human hair specimens has had legal precedence. The characterization and mapping of STR loci in domestic species, including cats and dogs, creates the potential to identify individual animals with high statistical confidence. We describe a homicide case where STR analysis of DNA extracted from the root of a cat hair linked to a crime scene in Prince Edward Island, Canada was compared to the STR profile of DNA from the pet cat of the primary suspect's family. The case set a legal precedent for the introduction of automated genotyping of pet animal hairs in forensic cases and stimulated the development of a formalized forensic typing system in domestic cats and population genetic database with which to compute match probabilities.

### TUESDAY 14:45 - 15:30 [P2 PLENARY] **Tools for comparative genomics** Frank Nicholas<sup>1,2,3,\*</sup> <sup>1</sup>Reprogen, Faculty of Veterinary Science, University of Sydney, NSW 2006 <sup>2</sup>CRC for Innovative Dairy Products <sup>3</sup>SheepGenomics **Keywords:** Comparative Genomics

The enormous global activity in genomics creates a challenge for the development of tools to exploit the rapid expansion of knowledge. A key challenge has been to integrate all available mapping information for a non-sequenced species into a single best-bet representation of the genome of that species. The genomes of non-sequenced and sequenced species can then be compared via Oxford grids, and virtual genome maps can be created for non-sequenced species. The recent development of locusorder maps derived solely from linkage disequilibrium data provides a new, powerful means for creating a map in species for which no conventional maps exist, and a resource for sequence assembly in species that have been sequenced.

#### THURSDAY 09:45 - 10:30 [P6 PLENARY]

# Wolbachia-mediated life shortening of Aedes aegypti to reduce dengue transmission ${\rm Scott}~{\rm O'Neill}^{1,*}$

<sup>1</sup>School of Integrative Biology, University of Queensland

Keywords: Pathogens, Parasites & Symbionts Pathogens, Parasites & Symbionts

Insect age is a critical factor influencing the dynamics of a range of insect transmitted diseases. Its importance is linked to the extrinsic incubation period (EIP) of a given pathogen within the vector. Since the EIP of many pathogens is quite long relative to the natural insect lifespan only old insects are of epidemiological importance. We have successfully transferred life-shortening Wolbachia symbionts into Aedes aegypti, the primary dengue vector, that halve adult lifespan in the laboratory. These bacteria are maternally inherited at high frequency and induce complete cytoplasmic incompatibility which may facilitate their natural spread into field populations. Interestingly the infection appears to influence the ability of these mosquitoes to successfully utilize the blood of certain hosts for egg production. Laboratory data suggest that this approach may be able to significantly reduce dengue transmission potential of Aedes populations. This potential will soon be evaluated in a field cage setting in Nth Queensland.

TUESDAY 09:00 - 09:45 [P1 PLENARY]

### Differentiation and development: The marsupial solution.

Marilyn Renfree<sup>1,\*</sup>

<sup>1</sup>University of Melbourne

**Keywords:** Comparative Genomics Developmental Genetics

Marsupials differ from eutherian mammals in their mode of reproduction. Their young are born at a very early stage of development and complete their growth in a pouch, or marsupium. They produce the smallest young, have the longest sperm, the longest period of embryonic diapause, the shortest pregnancy, become sexually differentiated after birth and have the most sophisticated lactation with a changing milk composition throughout the lengthy period of lactation of any mammal. Understanding the control of many of these processes is burgeoning. Embryonic diapause occurs in about 100 mammals, but the marsupial blastocyst enters diapause as a 100 unilaminar cell layer, whereas the eutherian blastocyst has an inner cell mass and thus the embryonic and placental tissues are already identifiable. Marsupials have a fully functional placenta that expresses imprinted genes as in eutherians, suggesting that genomic imprinting has a common origin in all therian mammals. In contrast to eutherians, some marsupial sexual dimorphisms are hormone independent, instead depending on a gene or genes on the X-chromosome. The neonatal marsupial is born with an immature immune system, so is an ideal recipient for xenografts. Testes grafted into neonatal females induce the recipient's ovaries to take on a testicular appearance, while testicular sex reversal can be achieved after administration of oestrogen to prematurely-born male neonates. Sexual differentiation occurs postnatally in distinct stages separated by wide windows of time, so is amenable to experimental manipulation. Early exposure to androgens hormonally imprints the brain and also the development of the prostate and phallus. Marsupials and eutherians diverged about 130 million years ago, but each developed a differing mode of reproduction. Current sequencing of the genome of two marsupial species will tell us much about the way in which these mammalian lineages evolved their separate, but equally successful reproductive strategies.

## FRIDAY 16:30 - 17:15 [P9 PLENARY]

Endosymbiotic evolution.

Jeremy N. Timmis

<sup>1</sup>Discipline of Genetics, School of Molecular and Biomedical Science, The University of Adelaide **Keywords:** Evolutionary Genetics Adaptation

Mitochondria and chloroplasts contain genomes which encode only a small fraction of the proteins required for their biogenesis and function. Most of the genes that were present in the prokaryotic ancestors of these organelles have been transferred to the nucleus during endosymbiotic evolution. Genes that relocated and became functional in the nucleus were deleted from the organelle genomes, reducing organelle genome size and abolishing mitochondrial and chloroplast autonomy. Early experiments, confirmed by genome sequencing, revealed many nuclear segments of extant mitochondrial and chloroplast DNA sequences. These range in size from small to very large, sometimes encompassing complete organelle genomes, integrated into the nuclear DNA of essentially all eukaryotes. These observations suggested that DNA transfer, and perhaps functional gene transfer, may be ongoing processes. Both these types of event have now been demonstrated at the molecular level. It is possible to monitor DNA transfer experimentally in some systems and the high frequencies observed indicate that the process is an ongoing natural mechanism that pervades nuclear DNA dynamics (Timmis et al., 2004). Though most of the organellar DNA integration events result in nonfunctional pseudogenes, the process of transfer and integration is clearly a prerequisite for functional gene relocation to the nucleus that has taken place in all eukaryotic lineages. Real time experiments in tobacco show that chloroplast DNA transposes to the nucleus at an astonishingly high frequency, suggesting that the process is capable of generating a high level of genetic novelty that has been a major driver of evolution that is unique to eukaryotes.

Timmis JN, Ayliffe MA, Huang CY, Martin W (2004) Endosymbiotic gene transfer: organelle genomes forge eukaryotic chromosomes. Nat. Rev. Genet., 5: 123-135



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# **Oral Abstracts** (in alphabetical order of first author)

### FRIDAY 15:30 - 15:45 [F4 CONSERVATION GENETICS 2] **Grey nurse shark genetics: what do we know and what do we need to know?** Heidi Ahonen<sup>1,\*</sup> Rob Harcourt<sup>1</sup> Adam Stow<sup>1</sup> <sup>1</sup>Macquarie University **Keywords:** Population Genetics Ancient and Forensic DNA

Anthropogenic impacts are believed to be the primary threats to the eastern Australian population of grey nurse sharks (*Carcharias taurus*), which is listed as Critically Endangered, and the most threatened population globally. Recently we have shown that grey nurse sharks sampled off eastern Australian are isolated and have low levels of genetic diversity However the degree of isolation between extant populations, and whether the low level of genetic diversity off the East coast is due to anthropogenic factors, is still uncertain. Therefore further sampling and genetic analysis throughout the global distribution of *C. taurus* are needed. In this study mitochondrial DNA (mtDNA) and nuclear loci (AFLP's and microsatellites) are being used to estimate genetic partitioning among grey nurse shark populations throughout their global distribution. Analysis of further mtDNA sequence data suggest less than a 5% possibility of finding an additional haplotype from the eastern Australia population. Given the longevity and life history of *C. taurus*, it is more likely that this depauperate genetic variation is due to historical rather than anthropogenic processes. To test this hypothesis and find out whether genetic variation was higher prior to anthropogenic impacts, DNA needs to be extracted from specimens from the 1970's or earlier, i.e. before the major population decline. For this purpose we have successfully extracted DNA from shark teeth and cartilage.

### THURSDAY 11:45 - 12:00 [TH1 POPULATION GENETICS 1]

# Oceanic variability and coastal topography shape local genetic structure in a longdispersing marine invertebrate

Sam Banks<sup>1,\*</sup> Maxine Piggott<sup>1</sup> Jane Williamson<sup>1</sup> Ulysse Bove<sup>1</sup> Neil Holbrook<sup>2</sup> Luciano Beheregaray<sup>1</sup> <sup>1</sup>Department of Biological Sciences, Macquarie University <sup>2</sup>Department of Physical Geography, Macquarie University

Keywords: Population Genetics Conservation Genetics

Understanding the scale of marine population connectivity is critical for the conservation and sustainable management of marine resources. For many marine species adults are benthic and relatively immobile, so patterns of larval dispersal and recruitment provide the key to understanding marine population connectivity. Contrary to previous expectations, recent studies have often detected unexpectedly low dispersal and fine-scale population structure in the sea, leading to a paradigm shift in how marine systems are viewed. Nonetheless, the link between fine-scale marine population structure and the underlying physical and biological processes has not been made.

To evaluate the influence of physical oceanographic, geographic and habitat-related variables on marine genetic structure, we genotyped microsatellite loci from populations of the broadcast-spawning sea urchin Centrostephanus rodgersii sampled from northern New South Wales to Tasmania and east to Lord Howe Island and New Zealand. Weak genetic differentiation and a lack of isolation-by-distance throughout the species' range were consistent with the long larval planktonic duration of C. rodgersii. However, we detected fine-scale patchy genetic structure, elevated intra-population genotypic autocorrelation and heterozygosity excess along the southeast Australian mainland coast. This pattern is consistent with a low Ne/N ratio resulting from high variance in individual reproductive success and was present in association with high inter-annual variability in sea surface temperature (SST) and coastal topographical complexity. The zone of high SST variability is characterised by periodic shedding of eddies from the East Australian Current, and we suggest that ocean current circulation may, through its influence on larval transport and recruitment, interact with the genetic consequences of large variance in individual reproductive success to generate patterns of fine-scale patchy genetic structure. If proven consistent across species, our findings suggest the optimal scale for fisheries management and reserve design should vary among localities in relation to regional oceanographic variability and coastal geography.

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# TUESDAY 11:15 - 11:30 [T1 COMPARATIVE GENOMICS 1]

A lousy genome! The 110 Mb genome of the human body louse, Pediculus humanus Ewen Kirkness1 Renfu Shao2 Stephen Barker<sup>2,\*</sup> for the Human Body Louse Genome Consortium <sup>1</sup>The Institute for Genomic Research, 9712 Medical Centre Drive, Rockville, MD 20850, U.S.A. <sup>2</sup>Parasitology Section, School of Molecular and Microbial Sciences, The University of Queensland Keywords: Comparative Genomics Comparative Genomics

The 110 Mb genome of the human body louse, Pediculus humanus, is small – much smaller than any sequenced insect genome, and similar to that of the nematode, C. elegans. The sequence of this compact genome can therefore help us identify novel functional elements among invertebrate genomes (cf fugu for vertebrate genomes). The human body louse has many additional interesting features, such as endosymbiotic bacteria, holocentric chromosomes, hemimetabolism (lack of pupal stage), and unusual mitochondrial DNA, that make it a very attractive candidate for genome analysis. It is also hoped that its genome sequence will enable novel approaches for controlling the incidence of this human parasite, and the diseases that it can transmit (epidemic typhus, relapsing fever and trench fever). The louse genome was sequenced to 8x coverage using a whole genome shotgun approach, with 1.2 million paired end reads from plasmid libraries. Few large insert fosmids or BACs were clonable, possibly due to the low GC content of the louse genome (28%). Despite the reliance on short insert clones, and substantial polymorphism among the hundreds of lice genomes that were pooled for sequencing, it was possible to generate an assembly with relatively long range continuity (scaffold N50 = 500 kb). The longest 300 scaffolds span more than 95% of the 111 Mb assembly. In the process of sequencing the louse genome, DNA from its principal bacterial endosymbiont (Candidatus Riesia pediculicola) was sequenced to ~50x coverage, and assembled to yield a single chromosome of 574 kb. An independent bacterial plasmid (6.5 kb) was also assembled. Annotation of the louse genome assembly has revealed a relatively uniform GC content across the genome, though introns are enriched for the CpG dinucleotide. Most of the repetitive DNA consists of simple repeats (~10% of genome), with few identifiable interspersed repeats (< 5%). Models for ~11,000 genes reveal dense clusters of compact genes with short introns. However, there are also large genomic spans where no genes have been identified, but which are likely to have functional significance. The chromosome of the bacterial endosymbiont contains ~600 genes, enriched for the biosynthesis of cofactors, and depleted for regulatory functions. This project provides the first comprehensive gene tally for an insect host and its bacterial endosymbiont, such that functional complementarity can be fully assessed.

### WEDNESDAY 12:15 - 12:30 [W1 COMPARATIVE GENOMICS 2]

Know your enemy – Detailed analysis of the the genome of the sheep blowfly, Lucilia cuprina

Philip Batterham<sup>1,\*</sup> Siu Fai Lee<sup>1</sup> Zhenzhong Chen<sup>1</sup> Alessandro Blasetti<sup>1</sup> Ayscha Hill-Williams<sup>1</sup> <sup>1</sup>SRC-CESAR, Bio21 Institute, University of Melbourne Keywords: Comparative Genomics Gene and QTL Mapping

Insecticides kill insects by interacting with a target protein that is required for survival. Resistance evolves due to mutations resulting in the modification or elimination of the target protein or enhanced detoxification of the insecticide. Traditional methods of insecticide discovery provide no information on the identity of target proteins. The capacity of insect enzymes to detoxify insecticides is poorly understood. When resistance does evolve the resistance gene can only be identified by a candidate gene approach because the poor molecular characterization of pest genome precludes positional cloning.

With funding from Australian Wool Innovation we have commenced a detailed analysis of the genome of the sheep blowfly, Lucilia cuprina. Our goal is to find genes that encode novel target proteins unique to the sheep blowfly, creating the potential for blowfly specific insecticides or vaccines to be designed. The isolation of genes encoding detoxification enzymes will allow the defence systems of the blowfly to be characterized in detail. We have end sequenced 30,000 cDNA clones. In the process we have identified up to 7464 different genes, corresponding to approximately 50% of the genes in the genome. The expression of these genes in different tissues and at different stages of the life cycle is currently being characterized using CombiMatrix Arrays.

Molecular genetic markers produced in this study will allow the structure and movement of blowfly populations to be monitored. Other genetic markers are currently being used to construct high resolution genetic maps of the genome. We have identified 952 L. cuprina genes that could serve as anchor loci, having a single clear orthologue in Drosophila melanogaster and the mosquito, Anopheles gambiae. The mapping of these genes by recombination has commenced. These maps will allow insecticide resistance genes to be isolated and identified.

## WEDNESDAY 11:15 - 11:30 [W2 PHYLOGEOGRAPHY]

# Mitochondrial and ribosomal DNA spacer evolution of the Southwest Pacific malaria vector Anopheles farauti s.s. reveals a complex demographic history and interesting concerted evolution.

Nigel W Beebe<sup>1,\*</sup> James E Bower<sup>2</sup> Robert D Cooper<sup>3</sup> <sup>1</sup>University of Technology Sydney <sup>2</sup>University of Wollongong <sup>3</sup>Australian Army Malaria Institute **Keywords:** Speciation and Phylogeography Population Genetics

Anopheles farauti s.s. is an important malaria vector of the southwest Pacific region (SWP) and one of 12 cryptic species in the An. punctulatus group. From collections spanning northern Australia, Papua New Guinea, the Solomon Islands and Vanuatu, we looked at the population genetic structure of this coastally restricted malaria vector using the mitochondrial cytochrome oxidase gene (COI) and the ribosomal DNA transcribed spacers 1 and 2 (ITS1 and ITS2). We found this mosquito-to be made up of several geographically and genetically structured populations with barriers to movement identified between islands as well as within the continents of Australia and Papua New Guinea. The concerted evolution if the ITS1 and ITS2 suggested substantial non-homogenised spacer variation fixed within populations and showed a much more rapid, but less quantitative, evolution than the COI.

### FRIDAY 11:00 - 11:15 [F1 POPULATION GENETICS 2]

**MEGMAR (Molecular Ecology Group for Marine Research): Understanding and predicting connectivity in the sea using codistributed population data sets** Luciano B Beheregaray<sup>1,\*</sup> Sam S Banks<sup>1</sup> Luciana M Moller<sup>1</sup> Maxine Piggott<sup>1</sup> Neil Holbrook<sup>2</sup> Shannon Corrigan<sup>1</sup> Joanna Wiszniewski<sup>1</sup> Kerstin Bilgmann<sup>1</sup> Kim Shaddick<sup>1</sup> Kathryn Newton<sup>1</sup>

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Keywords: Population Genetics Conservation Genetics

Systematic discussions about marine biodiversity usually converge on a single topic: the dispersal of marine organisms. Successful dispersal is perhaps the major process driving species distribution, persistence and evolution. Dispersal is also a critical factor when designing marine protected areas or developing management strategies for fishery and aquaculture resources. The Molecular Ecology Group for Marine Research (MEGMAR) is a new research initiative that uses powerful data and analytical tools in genetics and oceanography to understand how patterns of connectivity and diversity in the sea are generated and maintained. We have concentrated our efforts along the southeastern and southern Australian coasts, from where we amassed a large sample (84 sites, ~ 4,400 individuals) of dolphins (Tursiops spp. and Delphinus delphis), sharks (Orectolobus spp.), teleosts (Macquaria colonorum and M. novemaculeata), sea-urchins (Centrostephanus rodgersii), abalones (Haliotis coccoradiata), oysters (Saccostrea commercialis), and ascidians (Botrylloides leachii and Pyura stolonifera). We have developed microsatellite DNA markers for four of these taxa and generated and analysed multilocus DNA data for ~ 80% of our sample. Here we summarize results from our ongoing MEGMAR research program. We compare patterns of connectivity derived from genetic data in species with contrasting life histories and discuss our findings in relation to coastal oceanography and geomorphology. We show the value of a comparative approach for testing key hypotheses about marine connectivity and for identifying physical and biogeographic processes influencing marine biodiversity. This approach can potentially provide policy makers with a unique opportunity for incorporating information from key oceanographic factors into decision making strategies for reserve design and fisheries management.

### WEDNESDAY 12:45 - 13:00 [W2 PHYLOGEOGRAPHY]

# Fine-scale phylogeographic structure in an Amazonian flooded-forest ecosystem: a multispecies comparative study

Luciano B Beheregaray<sup>1,\*</sup> Ning L Chao<sup>2</sup> Maxine Piggott<sup>1</sup> Georgina Cooke<sup>1</sup> Shannon Corrigan<sup>1</sup> Aaron Harmer<sup>1</sup> Luciana Möller<sup>1</sup> Mark Sistrom<sup>1</sup> Joanna Wiszniewski<sup>1</sup> <sup>1</sup>Molecular Ecology Lab, Macquarie University <sup>2</sup>Universidade do Amazonas, Brazil **Keywords:** Speciation and Phylogeography Population Genetics

The richest biota in the world is found in the lowland rainforests of Amazonia. Despite their enormous importance as source of biodiversity, little is known about the evolutionary processes that generate diversification in Amazonia. We are using molecular approaches to elucidate the history of diversification of rainforest-dependent fishes from the Rio Negro floodplain (RNF), in central Amazonia. The RNF is home to an extraordinary diversity of small forest fish and is a major fishing

ground for live aquarium fish – an activity that accounts for about 60% of the income of RNF riverine communities. We have conducted extensive field expeditions and amassed samples of four fish groups (cardinal tetras, pencilfish, hatchetfish and rummy nose tetras) from 25 Rio Negro tributaries – a sampling effort that corresponds to 100 co-distributed populations and over 2,000 individuals. Here we summarize results for all species based on mtDNA and intron sequences and microsatellite DNA markers. Marked genetic differentiation between populations supports the proposal of a number of cryptic species in the RNF. Coalescence methods suggest a very complex history of diversification for phylogroups from the middle RNF, while marked levels of divergence between headwater populations appears as a result of isolation in disconnected tributaries. Analyses based on microsatellites confirm the general phylogeographic pattern for each species but also disclose significant genetic structure over micro-geographic scales (including absence of gene flow between populations separated by a few kilometers). We discuss on the implications of our comparative approach to evaluate the relative contribution of ecological and geographic processes underlying population differentiation and to test multiple hypotheses about speciation in tropical rainforests.

#### THURSDAY 11:00 - 11:15 [TH2 EVOLUTIONARY GENETICS]

### What Drives Evolution?

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<sup>1</sup>University of Melbourne

Keywords: Evolutionary Genetics Adaptation

The evolutionary synthesis of the 1940s 'combined Darwin's Natural Selection' with Genetics. The genetic subfields involved were population and quantitative genetics. These sciences developed mathematically what happens to genes in populations and in quantitative traits, starting with simple equations (e.g. P = G + E). The mathematics has been elaborated greatly. But the underlying thoughts (organisms change as a result of changing genes modified by environment) remain unchallenged as drivers of evolution.

I draw attention to flaws in quantitative and population genetics, which contradict the above explanation of evolution. The flaws include:

- a. Falconer's textbook of Quantitative Genetics in all editions states that 'natural selection was assumed to be absent' in the development of the subject. Hence current quantitative genetics cannot describe natural selection.
- b. All organisms are limited by the resources of their environment (more and better grass feeds more and better herbivores). Nature selects those organisms using environmental resources most efficiently.
- c. In a stable environmental niche, natural selection can do no more than adapt animals perfectly to the resources and challenges of the environment. Organisms other than those optimal for that environment are continually culled. Thus for most of evolutionary time natural selection has prevented changes.
- d. The fossil record shows very rapid evolution after extinctions, often after global calamities. Evolutionary changes occur when resource become available to survivors after other species died out.

The facts b), c) and d), each taken for granted in its subfield of evolution, have not been put together with the evolutionary synthesis. When they are, the following explanation becomes obvious: Environmental change selects organisms able to use the changed environments efficiently. Change usually follows extinctions for whatever reason of organisms previously adapted to the niche. Genes prevent selected phenotypic changes dissipating again. Selection of genes in a gene pool is a metaphor that has misled evolutionary biology for over 60 years. Putting aside this metaphor will greatly affect how we see many aspects of genetics.

#### FRIDAY 12:45 - 13:00 [F2 CONSERVATION GENETICS 1]

### **Devil Facial Tumour Disease: cytogenetic clues to transmission and development** Hannah Sarah Bender<sup>1,\*</sup> Amber Alsop<sup>1</sup> Anne-Maree Pearse<sup>2</sup> Jenny AM Graves<sup>1</sup>

<sup>1</sup>Comparative Genomics Group, Research School of Biological Sciences, The Australian National University, Canberra, ACT 2601

<sup>2</sup>Department of Primary Industries and Water, Animal Health Laboratory, Kings Meadows TAS 7249 **Keywords:** Comparative Genomics Conservation Genetics

Devil Facial Tumour Disease (DFTD) is a contagious tumour associated with a devastating decline in wild Tasmanian devil (*Sarcophilus harrisii*) populations over the past ten years. Lesions are initially small nodules arising in the dermis of the oral cavity, head or neck. These rapidly progress to large, ulcerated, masses, obstructing affected animals from feeding and resulting in death, usually within five

months of the lesion first appearing. The allograft hypothesis of transmission (Pearse and Swift, Nature, 2006) arose from the extraordinary discovery that the complex chromosome rearrangements found in the tumour cells are identical between unrelated animals. The theory proposes that the cancer cells themselves are the infective agent, transmitted between animals by biting, a concept with fascinating implications for transplant medicine and our understanding of tumour development and progression. We are using chromosome painting to confirm the apparent similarity of the tumour karyotypes from different devils in order to test the hypothesis. Tumour cells have been cultured and harvested from numerous tumour biopsies collected in the field. Normal chromosome paints created from flowed-sorted devil chromosomes are being hybridised to metaphase preparations from tumour cultures to reveal which portions of the normal karyotype have been rearranged in the tumours. Preliminary results indicate that the tumour karyotype is more complex than G-banding analysis would suggest and that the rearrangements are indeed identical between animals. We are currently isolating individual tumour chromosomes by microdissection in order to generate tumour-specific paints using degenerate oligonucleotide-primed PCR. By applying tumour paints to normal metaphases (reverse painting) we will be able to pinpoint chromosome breakpoints and identify regions of the genome involved in carcinogenesis.

### FRIDAY 15:45 - 16:00 [F4 CONSERVATION GENETICS 2]

# Marked genetic differentiation in short-beaked common dolphins subject to fisheries impacts in southern Australia

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Keywords: Conservation Genetics Population Genetics

Interactions between short-beaked common dolphins (Delphinus delphis) and the fishing industry of South Australia have lead to serious concerns over the long term viability of the local dolphin population(s). Common dolphins are gregarious animals that show high vagility and are therefore generally expected to display reduced genetic differentiation over large spatial scales. Here we investigate the population genetic structure of southern Australian common dolphins using data from mitochondrial DNA and microsatellite markers. Our aim is to identify management units (MUs) for conservation of the potentially threatened population(s). Our results showed no genetic structure in common dolphins along the coast of South Australia. By contrast, we detected marked differentiation between dolphins from South Australia and southeastern Tasmania, suggesting a minimum of two populations in southern Australia. We hypothesise that the ephemeral distribution of the common dolphins' main prey item in South Australia - small pelagic fish - enhances movement and dispersal between dolphin groups. On the other hand, clear differences in water temperature, habitat features and fish abundance between South Australia and southeastern Tasmania may have contributed to the partial isolation of these dolphin populations. The pattern of differentiation detected here is remarkable compared to the low levels of genetic structure reported elsewhere for this species. Considering that South Australia has the largest volume purse-seine fishery in Australia and that this is known to causes fatal interactions of common dolphins every year, it is critical to manage the two dolphin populations separately. Recommendations for assessing the impacts of the fishery are presented.

### TUESDAY 18:05 - 18:25 [P3 CATCHESIDE PRIZE]

# An ENU screen for modifiers of epigenetic reprogramming reveals novel sex-specific and parental effects

Marnie Blewitt<sup>1,2,\*</sup> Nicola Vickaryous<sup>1,2</sup> Alyson Ashe<sup>1,3</sup> Sarah Hemley<sup>1</sup> Jost Preis<sup>1</sup> Ruth Arkell<sup>4</sup> Emma Whitelaw<sup>1,3</sup>

<sup>1</sup>University of Sydney, <sup>2</sup>Walter and Eliza Hall Institute for Medical Research, <sup>3</sup>Queensland Institute of Medical Research, <sup>4</sup>Mammalian Genetics Unit, MRC Harwell **Keywords:** Epigenetics Functional Genomics

Epigenetic reprogramming of the genome is an essential process, which occurs during both primordial germ cell development, and early embryogenesis, so that the epigenetic marks are cleared and reset between generations. Occasionally, it appears that this epigenetic reprogramming is incomplete, since we observe transgenerational epigenetic inheritance. Non-Mendelian inheritance, such as this, changes the way we think about the inheritance of phenotypic traits.

We have established a sensitised screen to identify genes involved in epigenetic reprogramming, using ENU mutagenesis of a line of mice carrying a variegating transgene. The mutagenesis screen has produced 13 mutant lines (now at 17), six of which were identified in a screen for dominant mutations. All of these are semi-dominant and show some degree of homozygous embryonic lethality. The viability of the mutants, often depends on sex. For example, homozygous females of one of these mutations, die at mid-gestation, and do not appear to undergo normal X-inactivation. Linkage analysis reveals that they map to unique chromosomal locations, and many of the point mutations have now been identified.

Many of the dominant mutations affect epigenetic inheritance at the Avy allele. In all cases, we see sexspecific effects. The behaviour of the mutant lines suggests a common mechanism between Xinactivation, and transgene and retrotransposon silencing. Moreover, these mutations display both maternal and paternal effects, ie, heterozygosity in either parent affects the penetrance of wild-type offspring. This is a curious effect with no mammalian precedent, suggesting that the corresponding proteins are involved in chromatin packaging in the gametes, which affects gene expression in the subsequent offspring.

### FRIDAY 15:15 - 15:30 [F5 ADAPTATION]

### **LDH-B** and temperature tolerance in coral trout (Plectropomus leopardus) Emmanuelle Botte<sup>1,\*</sup>

<sup>1</sup>School of Marine and Tropical Biology, James Cook University, Townsville, QLD 4811, Australia. **Keywords:** Adaptation Evolutionary Genetics

With predicted increases in ocean temperatures associated with global warming it is essential that we understand how fish respond to temperature. Most research to date has studied acclimation and adaptation of temperate fish, despite the fact that tropical waters already experience more rapid temperature increases than temperate waters. Therefore, it is crucial that we investigate temperature tolerance in tropical fish so that we can predict how they will respond to increases in temperature. As a novel approach, we are using molecular tools to understand the mechanisms underlying adaptation and acclimation of coral trout (Plectropomus leopardus) to temperature.

Our study uses two genetically distinct populations of coral trout from different thermal environments (tropical vs sub-tropical). The aim of the study is to assess how expression of the candidate gene lactate dehydrogenase B (LDH-B) is influenced by different thermal regimes and whether there is evidence for thermal adaptation or acclimation at this gene that allows fish to cope with the differing temperatures.

In this study, quantitative PCR will be used to identify LDH-B expression level differences between fish from the two populations when subjected to cool, medium or warm experimental temperatures. If differences are observed, the underlying molecular mechanisms will be investigated by constructing and screening a genomic library to allow complete characterization of the LDH-B 5' and 3' UTRs. Sequence variation will be examined between the two populations to identify polymorphisms that might be present and responsible for changes in gene expression. This study will be one of the first to examine the molecular mechanisms behind thermal adaptation and acclimation in a tropical fish and will increase our understanding of physiological processes involved in temperature tolerance.

## THURSDAY 12:30 - 12:45 [TH3 FUNCTIONAL GENOMICS 2]

Moderate effects of myostatin F94L on cattle carcass traits

G S Sellick1 W S Pitchford1 C A Morris<sup>2</sup> N G Cullen<sup>2</sup> A M Crawford<sup>2</sup> H W Raadsma<sup>3</sup> C D.K. Bottema<sup>1,\*</sup>

<sup>1</sup>Livestock Systems Alliance, University of Adelaide <sup>2</sup>AgResearch <sup>3</sup>Reprogen, University of Sydney **Keywords:** Comparative Genomics Gene and QTL Mapping

In a cattle gene mapping project involving a Limousin x Jersey backcross, a highly significant quantitative trait locus (QTL) was found at 0-15 cM on bovine chromosome 2 for meat percent, eye muscle area and silverside percent. In this region, there was a strong positional candidate gene, myostatin (MSTN or growth differentiation factor 8, GDF8), as loss-of-function mutations in the myostatin gene cause an extreme "double muscling" phenotype in cattle. Linkage disequilibrium analysis of the maternally inherited myostatin haplotypes from outbred Limousin and Jersey cattle provided overwhelming support for highly significant associations of myostatin with cattle carcass traits. Although there were many polymorphisms discovered in the myostatin gene, there was just one potentially functional polymorphism identified by sequencing. This polymorphism was a "C" to "A"

transversion in the mRNA (GDF8g.433C>A), which results in the amino acid substitution of phenylalanine94 by leucine in the inhibitory domain of the protein. The amino acid substitution was the only polymorphism consistently related to increased muscling. Overall, the size of the SNP g.433C>A additive effect on cattle carcass traits was moderately large with the "A" allele found to be associated with a 5.5% increase in silverside percent and eye muscle area and a 2.3% increase in total meat percent relative to the "C" allele. These phenotypic effects of the "A" allele were partially recessive. Although the SNP g.433C>A had been identified previously, the phenotypic effects of this DNA variant have not been recognized. In sheep, an illegitimate microRNA site in the 3'UTR of the myostatin gene has been found to down-regulate translation and hence, increase muscling in the Texel breed. No difference in gene expression of the myostatin gene was observed herein. The present study provides strong evidence that an amino acid substitution in myostatin can also produce an intermediate, non-doubling muscling phenotype.

WEDNESDAY 14:15 - 14:30 [W6 FUNCTIONAL GENOMICS 1]

Dominance in absentia - phantom rec+ genes. Frederick J Bowring<sup>1,\*</sup> P Jane Yeadon<sup>1</sup> David E A Catcheside<sup>1</sup> School of Biological Sciences, Flinders University Keywords: Functional Genomics RNAi and non-coding

DNA rec 1+, rec 2+ and rec 3+ are dominant trans-acting genes that suppress meiotic recombination in specific regions of the Neurospora crassa genome. As an example, up to 1% of progeny from a rec 2 by rec 2 cross experience recombination at his 3 but this falls to about 0.005% when one or both parents carries rec 2+. Our attempt to clone rec 2+ has encountered substantial difficulty and the main reason for this has recently become obvious - rec 2+ does not exist!

Approximately 10kb of the rec 2+ chromosome is replaced with a unique 3kb stretch in rec 2 strains. However, rec-2+ DNA seems to lack function since putting rec 2+ sequence into rec 2 strains fails to yield a rec 2+ phenotype while deletion of rec-2+ sequence does not give a rec-2 phenotype. With the discovery of meiotic silencing by unpaired DNA (MSUD), we wondered whether rec 2+ might simply appear dominant in rec 2+/rec 2 heterozygotes because it deprives rec 2 of an opportunity to pair during meiosis. This appears to be the case.

Disabling MSUD in rec 2+/rec 2 heterozygotes substantially increases recombination at his 3. Similarly, in the absence of MSUD, the amount of recombination at his 1 in rec 1+/rec 1 heterozygotes and at am in rec-3+/rec-3 heterozygotes is indistinguishable from that in rec 1 and rec-3 homozygotes respectively. Thus, rather than rec+ genes producing suppressors of recombination it now appears likely that the products of rec 1, rec 2 and possibly rec 3 act to promote recombination.

#### WEDNESDAY 12:15 - 12:30 [W2 PHYLOGEOGRAPHY]

# Using ancient DNA to assess the phylogeography of European, Russian and North American brown bears (Ursus arctos). Sarah Bray<sup>1,\*</sup> Jeremy Austin<sup>1</sup> Alan Cooper<sup>1</sup>

<sup>1</sup>University of Adelaide

Keywords: Ancient and Forensic DNA Phylogeny, Biodiversity & Barcoding

Although modern genetic data is commonly used to assess the phylogeography and evolutionary history of extant species and populations, the study of ancient DNA provides us with a different view which allows us to trace historic and ancient gene flow through time in response to environmental change. Brown bear specimens are widely distributed across the Northern Hemisphere through both space and time and have been shown to exhibit pronounced mitochondrial phylogeographic structure, making it an ideal species to investigate the effects of climate change and human impact on genetic diversity. The phylogeography of modern brown bears has been relatively well studied, however little molecular data exists for Holocene or late Pleistocene material (<50 000 years) across Europe and Russia. We have used ancient DNA techniques to retrieve mitochondrial DNA sequences from Holocene and late Pleistocene brown bears across North America, Russia and Europe. This data will be used to investigate the genetic diversity and phylogeographic history of brown bears across the Northern Hemisphere. Preliminary results suggest that the modern contact zone between brown bear clades in Scandinavia has existed for at least the last 6000 years, and that a similar contact zone existed through the Holocene period in Austria. Parallel studies on the New World hypercarnivorous Tremarctine bears have revealed surprising phylogenetic relationships.

#### WEDNESDAY 15:30 - 15:45 [W5 PHYLOGENY]

#### Evolutionary conservation of microsatellites in mammalian genomes

Emmanuel Buschiazzo<sup>1,\*</sup> Neil Gemmell<sup>1</sup>

<sup>1</sup>University of Canterbury, Christchurch, New Zealand

Keywords: Evolutionary Genetics Population Genetics Gene and QTL Mapping Speciation and Phylogeography

Microsatellites are among the most versatile of genetic markers, being used in an impressive number of biological applications. However, the evolutionary dynamics of these markers and their function in genomes remain a source of contention. Almost 20 years after the discovery of these ubiquitous simple sequences, new genomic data are clarifying our understanding of the structure, distribution and variability of microsatellites in genomes, especially for the eukaryotes. We have recently reviewed(1) the mutational processes, biases and constraints believed to be involved in the evolution of microsatellites, particularly with respect to the creation and degeneration of microsatellites, which can be broadly viewed as a life cycle. We propose to test this concept in mammals using a blend of bioinformatics and comparative analysis above the species level. Our preliminary results unveil the unexpected extent of the conservation of microsatellite loci across mammalian species. We believe that analyses of variability of these conserved loci at the population level will provide invaluable information on the evolutionary course of microsatellites in eukaryotes and will be the ground for the development of a realistic model of evolution at these loci. 1. Buschiazzo, E and Gemmell, NJ (2006). The rise, fall and renaissance of microsatellites in eukaryotic genomes. *BioEssays* 28: 1040-1050.

#### FRIDAY 14:15 - 14:30 [F4 CONSERVATION GENETICS 2]

### Pollen dispersal between planted populations and remnant native populations in a fragmented agricultural landscape

Margaret Byrne<sup>7,\*</sup> Jane Sampson<sup>1</sup> Melissa Millar<sup>2</sup> <sup>1</sup>Science Division, Department of Environment and Conservation <sup>2</sup>Faculty of Sciences, The University of Adelaide **Keywords:** Population Genetics Conservation Genetics

Planting of native species is becoming extensive in fragmented agricultural landscapes in both rehabilitation and restoration programs. It is important to have knowledge of patterns of gene flow since it may be beneficial where the objective is to restore native populations and landscape processes, or may be undesirable if particular plantings are undertaken for more specific objectives such as hydrological management. Eucalyptus loxophleba and Acacia saligna are two woody species being planted in agroforestry programs to address amelioration of dryland salinity in the agricultural region of Western Australia. Both species have widespread distributions in Western Australia and their morphological and genetic variation is represented in subspecies. Planting of one subspecies within the natural range of another may lead to genetic contamination of natural remnant populations if pollen dispersal is extensive and the subspecies are inter-fertile. Pollen dispersal between planted and native remnant populations was investigated in both species where they occur in a mosaic of agricultural production. Extensive levels of long distance pollen dispersal were observed in both species over distances of up to 2 km. In addition the significantly greater fecundity of the planted subspecies of A. saligna lead to high levels of pollen immigration in the less fecund natural subspecies. The extensive pollen dispersal observed in these two species indicates that the risk of genetic contamination of remnant populations is significant. A risk management framework is required to ensure that agroforestry programs can be developed to achieve rehabilitation outcomes without negative impacts on remnant patches of biodiversity.

#### FRIDAY 11:45 - 12:00 [F2 CONSERVATION GENETICS 1]

### Genetic variation in translocated northern quoll (Dasyurus hallucatus) island populations.

Maria J Cardoso<sup>1,\*</sup> Brooke Rankmore<sup>2</sup> William B Sherwin<sup>1</sup> Karen B Firestone<sup>1</sup> <sup>1</sup>Department of Biological, Earth and Environmental Sciences, University of NSW <sup>2</sup>Department of Natural Resources, Environment and the Arts, Darwin, NT **Keywords:** Conservation Genetics Population Genetics

The northern quoll (Dasyurus hallucatus) is the smallest of the Australian quoll species. It is a solitary, nocturnal and largely insectivorous carnivore, though as an opportunist, its diet may also include small mammals, amphibians, reptiles and plants. The geographical distribution of the northern quoll has been reduced to six isolated regions in northern Australia. Threats to its survival include habitat destruction, changed fire regimes and the invasion of the cane toad (Bufo marinus) into areas of

its range. To this effect, the northern quoll is now listed as endangered under the current Commonwealth Environment Protection and Biodiversity Act. In 2003, as a form of insurance policy against extinction, northern quolls captured from mainland Northern Territory populations were translocated to two invasive animal-free offshore islands. These island populations are being monitored each year, with samples taken for genetic analysis. Genotypes at six polymorphic microsatellite loci revealed slightly lower genetic variation in the translocated populations (He 0.610-0.639; AR 3.33-3.37) compared to mainland populations (He 0.623-0.646; AR 3.61-3.79), but higher than that found in other Northern Territory islands (He 0.129-0.396; AR 1.34-2.33) that may have gone through past bottlenecks and/or long-term isolation. There is also evidence of low, but highly significant (p = 0.001) genetic differentiation between the translocated populations and the source mainland population, possibly due to factors underlying local adaptation and genetic drift that are often common in closed island populations. To ensure that these populations remain viable and are able to successfully adapt to changing environmental conditions, they should continue to be monitored so that effects associated with genetic erosion may be readily detected over time, thus allowing rapid and well informed management decisions to be made accordingly.

#### FRIDAY 11:30 - 11:45 [F1 POPULATION GENETICS 2]

An Update on Inbreeding in Australian Thoroughbred Racehorses Kao  ${\rm Castle}^{{\rm I},*}$ 

<sup>1</sup>Reprogen, Faculty of Veterinary Science, University of Sydney, NSW 2006 **Keywords:** Population Genetics

Recent Australian Thoroughbred breeding has seen a significant reduction in sire numbers from a peak of 2477 in 1987 to 836 in 2005. The number of foals born each year has steadied since a population growth period in the 1970s and 1980s, and has remained constant at around 16,500 to 18,000 per annum for the last fifteen years.

The use of shuttle stallions (stallions from overseas that serve mares in both hemispheres) began in 1988. The number of foals sired by shuttle stallions has grown consistently since the practice was introduced. Shuttle stallions now produce almost one third of the total Thoroughbred foal population in Australia each year, although they comprise only 8% of the total number of stallions at stud. The Australian Thoroughbred breeding population has included a significant proportion of imported horses even prior to the advent of shuttle stallions. In 1985, almost 49% of registered Thoroughbred foals had a sire or dam that was born overseas. In 2005, this proportion had increased to over 58%.

Inbreeding coefficients for 364,995 Australian Thoroughbreds born between 1985 and 2005 were calculated from all available data (an average of 22.8 generations of pedigree, comprising 799,734 animals in total). The average inbreeding coefficient for Australian Thoroughbreds born in 2005 was 0.1433, with a standard deviation of  $\pm 0.0104$ . The average rate of inbreeding over the past decade has been 0.00027 per year. This relatively low rate of inbreeding suggests that the transition to a smaller group of sires, combined with increased use of imported breeding stock, has not posed a threat to the genetic well-being of the Australian Thoroughbred.

#### THURSDAY 11:30 - 11:45 [TH1 POPULATION GENETICS 1]

#### **Racial origin of commercial and feral honey bees (Apis mellifera) in Western Australia** Nadine C Chapman<sup>1,\*</sup> Ben P Oldroyd1

<sup>1</sup>Behaviour and Genetics of Social Insects Laboratory, School of Biological Sciences, University of Sydney

#### Keywords: Population Genetics

Due to the introduction of exotic bee diseases in the eastern states, the Western Austalian boarders were closed to the import of bees for breeding and other purposes over 25 years ago. To provide genetically improved stock for the industry, a closed-population breeding program was established which now provides stock for the majority of Western Australian beekeepers. Given concerns that inbreeding would result from the closed population, we were asked to assess the genetic relatedness of WA breeding stocks using microsatellite and mitochondrial markers. We found that the breeding population still maintains considerable genetic diversity. The relatedness of the commercial bees to the feral Western Australian population was also investigated. We found that the feral population was genetically distinct from the genetic stock of the breeding program, but not from the genetic stock of beekeepers outside of the program. The honey bees of Western Australia are of two subspecies and three mitotypes, A. m. ligustica (mitotypes C1 and M7b) and A. m. iberica (mitotype M6). All but one commercial sample was of the preferred beekeeping subspecies A. m. ligustica. The feral population consists of both subspecies.

#### WEDNESDAY 11:30 - 11:45 [W3 BIOINFORMATICS] Effects of sampling and model parameters in cophylogenetic analysis Michael Charleston<sup>1,\*</sup> Michael Huang1 <sup>1</sup>University of Sydney Keywords: Phylogeny, Biodiversity & Barcoding Adaptation

Cophylogenetic analysis - the study of how groups of ecologically linked species such as parasites and their hosts were related in the past, based on their estimated phylogenies - is fraught with statistical challenges. Not least of these is that the number of possible solutions to a given instance of the problem grows exponentially with the number of hosts, the number of parasites, and with the degree of disagreement between the phylogenies. Further, the relative frequencies of the four recoverable largescale coevolutionary events of codivergence, duplication, host-switching and loss are very hard to estimate. Rather than force arbitrary values on these event types we can create a set of Pareto-optimal solutions, which are those that are in a sense "potentially" the best solutions, in that any one of them could have a minimal score for a particular feasible set of event costs. The number of Pareto-optimal solutions grows less rapidly than does the number of all possible solutions but is still large, so any estimation of the event costs values that could help reduce the dimensionality and therefore the size of the Pareto set would be a significant improvement. Another difficulty arises from the reasonable approach to taxon sampling, of once having identified a particular parasite on a particular host, to stop looking elsewhere, under the assumption that parasites are host-specific. This often means that some taxa are better sampled than others, but the effect of sampling error on cophylogenetic reconstruction is unknown: how does the choice of which taxa are chosen (or missed) affect our inferences? This talk describes some first steps in the estimation of model parameters that correspond to real instances in cophylogenetics and shows how they may be used to to improve our attempts to recover ancient history of associations between linked organisms.

#### THURSDAY 12:15 - 12:30 [TH3 FUNCTIONAL GENOMICS 2]

### A novel role for Xist RNA in the formation of a repressive nuclear compartment into which genes are recruited when silenced.

Julie Chaumeil<sup>1,2,\*</sup> Patricia Le Baccon<sup>1</sup> Anton Wutz<sup>3</sup> Edith Heard<sup>1</sup> <sup>1</sup>Mammalian Developmental Epigenetics Group - Curie Institute - Paris - France <sup>2</sup>Current Address: Comparative Genomics Group - RSBS - ANU - Canberra <sup>3</sup>Mammalian X-chromosome Inactivation Group - IMP - Vienna - Austria **Keywords:** Epigenetics

X chromosome inactivation is the mechanism by which dosage compensation for sex chromosomes between males and females is achieved in mammals. It represents a remarkable example of epigenetic gene regulation during early female development, as one of the two homologous X chromosomes is stably silenced and acquires some heterochromatin characteristics. Coating of the X chromosome by the non-coding Xist RNA is essential for the initiation and propagation of X inactivation. However, little is known about the mechanisms that transform this signal into silencing. Using mice female embryonic stem cells as a model system, as in vitro differentiation is accompanied by X inactivation, we investigated the early events underlying X inactivation. We showed that exclusion of RNA Polymerase II and transcription factors from the Xist RNA-coated X chromosome represents the earliest event following Xist RNA accumulation described so far in differentiating ES cells. Paradoxically, exclusion of the transcription machinery occurs before gene silencing is complete. However, examination of the 3D organization of X-linked genes revealed that when transcribed, they are always located at the periphery of, or outside, the Xist RNA domain, in contact with the transcription machinery. Upon silencing, genes shift to a more internal location, within the Xist RNA compartment devoid of transcription factors. Surprisingly, the appearance of this compartment is not dependent on the A-repeats of the Xist transcript, which are essential for gene silencing. However, the A-repeats are required for the relocation of genes into the Xist RNA silent domain. Thus, we propose that Xist RNA has multiple functions : A-repeat independent creation of a transcriptionally silent nuclear compartment; and A-repeat dependent induction of gene repression, which is associated with their translocation into this silent domain.

#### FRIDAY 12:45 - 13:00 [F3 MAPPING]

Homozygosity mapping of an autosomal-recessive Rosai-Dorfman-like condition in Lebanese-Australian families using high-density SNP genotyping.

Simon T Cliffe<sup>1,\*</sup> Tony Roscioli<sup>1</sup> Melanie Wong<sup>1</sup> Artur Darmanian<sup>1</sup> Greg Peters<sup>1</sup> Michael F Buckley<sup>1</sup> Robert Lindeman<sup>1</sup>

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Keywords: Gene and QTL Mapping Gene and QTL Mapping

Homozygosity mapping is a technique used to identify the genetic basis of autosomal-recessive diseases in consanguineous families. Traditionally STR markers have been used to detect regions of homozygosity, however high-density SNP genotyping is becoming more prevalent due to advantages in both cost and resolution. The binary nature of SNP alleles accords low marker informativeness, which is overcome by using a much greater density of markers than a typical STR marker study. This can provide a greater resolution, but requires a higher degree of analysis to detect significant regions of identity. This study outlines the analysis procedure used to examine 55, 721 SNP genotypes in two consanguineous Lebanese-Australian families with an autosomal-recessive Rosai-Dorfman-like condition.

#### FRIDAY 14:30 - 14:45 [F4 CONSERVATION GENETICS 2]

# Plant Mating Systems And Assessing Population Persistence In Fragmented Landscapes

David Coates<sup>1,\*</sup> Colin Yates<sup>1</sup> Jane Sampson<sup>1</sup> <sup>1</sup>Department of Environment and Conservation, Western Australia **Keywords:** Conservation Genetics Population Genetics

Population size and habitat disturbance are key factors likely to shape the mating system of populations in disturbed and fragmented landscapes. They would be expected to influence pollinator availability and behaviour, the ability to find mates in self incompatible species, inbreeding in self compatible species and the size of the pollen pool. These in turn might be expected to influence key variables critical for population persistence such as seed production, seed germination and seedling fitness. Here we investigate mating system variation in six rare species: Banksia cuneata, Banksia oligantha, Lambertia orbifolia (Proteaceae); Verticordia fimbrilepis subsp. fimbrilepis, Eucalyptus rameliana (Myrtaceae); Acacia sciophanes (Mimosaceae) and two common species Calothamnus quadrifidus (Myrtaceae) and Acacia anfractuosa. All seven species are animal pollinated relatively long lived woody shrubs with mixed mating systems. Population variation in mating system parameters was investigated in relation to population size and habitat disturbance. We show that although the mating system will vary depending upon pollination biology and life -history, as populations get smaller and habitat disturbance increases there is a trend towards increased inbreeding, smaller effective sizes of paternal pollen pools and greater variation in outcrossing among plants. From the species investigated in this study we have found that changes in the mating system can be useful indicators of population processes and can give valuable insight into the development of conservation strategies for plant species persistence following anthropogenic disturbance and landscape fragmentation.

#### WEDNESDAY 11:30 - 11:45 [W2 PHYLOGEOGRAPHY]

**Phylogeography of marine and brackish water Mollusca in southeastern Australia** Don Colgan<sup>1,\*</sup> Pam Da Costa<sup>1</sup> Tina Reutelshoefer<sup>1</sup>

<sup>1</sup>Australian Museum

Keywords: Speciation and Phylogeography Evolutionary Genetics

Each of the few studies of the phylogeography of southeastern Australian marine and brackish water environments has revealed further complexity. The complexity is likely due to the interaction of multiple factors including (i) the repeated challenges posed to fauna by environmental change in the Quaternary; (ii) intra-specific reproductive characteristics and responses to selection; and (iii) sporadic long-distance dispersal. A comparative understanding of gene flow in the region is important for management of MPAs. What is already known indicates, however, that much more research is required. Several studies of marine species have suggested that land-bridges across Bass Strait have had a prominent role in structuring genetic diversity in the region, but with conflicting inferences about timing. No study has closely identified the other principal biogeographic boundary in southern NSW with phylogeographic discontinuities. Studies of segments of the mitochondrial 12S rRNA and nuclear ITS-1 genes of the two nominal species of the estuarine/lagoonal hydrobiid snail *Tatea* have revealed new diversity patterns. In 12S rRNA, the same common haplotype is found in both *T. kesteveni* and *T. huonensis*, ranging from Ballina in northern NSW to Perth, WA. Variant haplotypes differ from the common form at few bases and have restricted (although occasionally disjunct) distributions. In ITS-1, the most frequent haplotype is widely-distributed in both species and a second common wide-ranging type is found in *T. kesteveni*.

We will report the results of investigations of other estuarine species (including the pulmonate snails *Phallomedusa* and *Salinator*, and the mussel *Xenostrobus securis*) to determine whether the Tatea pattern is general in molluscs from this environment. We will also report on studies searching for novel phylogeographic breaks in marine species such as the mussels *X. pulex* and *Brachidontes rostratus* and the snails *Siphonaria* spp., *Austrocochlea* spp. and *Bembicium* spp.

#### FRIDAY 12:00 - 12:15 [F3 MAPPING]

Tammar Wallaby Genetic Resources

Des Cooper<sup>1,\*</sup> Cathy Herbert<sup>1</sup> Kyall Zenger<sup>2</sup> Marilyn Renfree<sup>3</sup> Jenny Graves<sup>4</sup> Sue Forrest<sup>5</sup> <sup>1</sup>University of New South Wales, <sup>2</sup>University of Sydney, <sup>3</sup>University of Melbourne, <sup>4</sup>Australian National University, <sup>5</sup>WEHI

Keywords: Comparative Genomics Gene and QTL Mapping

The tammar wallaby (Macropus eugenii) is the "model" Australian marsupial species. Its reproductive biology has been investigated in great detail, because it possesses the phenomenon of embryonic diapause which is mediated by lactation in the first half of the year and by photoperiod in the second half. Its genome is being sequenced. The relationship of its karyotype to that of other mammals is being defined by chromosome painting. A linkage map is being developed in considerable detail. Two main subspecies exist, which can be crossed and then backcrossed. The South Australian subspecies is represented chiefly by the Kangaroo Island population (M. e. decres), which is very numerous. The mainland South Australian form (M. e. eugenii) survives only in New Zealand to which it was introduced in the 19th Century. The Western Australian subspecies (M. e. derbianus) still exists in small numbers on the mainland. Island populations are to be found in the Recherche Archipelago, Houtman Abrolhos Archipelago and on Garden Island. These populations represent sources of genetic variation, which may be useful for further research.

#### THURSDAY 12:45 - 13:00 [TH1 POPULATION GENETICS 1]

Immunocontraception of mammalian wildlife: ecological and immunogenetic issues Desmond W Cooper<sup>1,\*</sup> Larsen Elisabeth<sup>1</sup>

<sup>1</sup>The University of New South Wales

Keywords: Population Genetics Evolutionary Genetics

Immunocontraception involves stimulating immune responses against gametes or reproductive hormones thus preventing conception. The method is being developed for the humane control of pest and overabundant populations of mammalian wildlife. This paper examines three fundamental issues associated with its use: (1) the difficulties of obtaining responses to self-antigens, (2) the likely evolution of genetically based non-response to immunocontraceptive agents, and (3) the possible changes in the array of pathogens possessed by the target species after generations of immunocontraceptive are at present very basic. Should they be overcome, the effects of immunocontraception on the immunogenetic constitution of wildlife populations through selection for nonresponders must be examined. We suggest that the attempt to use the animal's own immune system to modulate reproduction may be incompatible with the basic biological function of protection against infectious disease. Research programs on mammalian immunocontraception should involve measurement of the heritability of non response and an assessment of the likely change in the response of the contracepted population to possible pathogens.

This work has been published in Reproduction (2006) 132(6):821-8

#### THURSDAY 11:15 - 11:30 [TH3 FUNCTIONAL GENOMICS 2]

A potent antimicrobial protein expressed in wallaby milk

Jianghui Wang<sup>1</sup> Benjamin G Cox<sup>1,\*</sup> Jane C Whitely<sup>1</sup>

Department of Primary Industries, 475 Mickleham Rd, Attwood, Victoria, Australia. Keywords: Functional Genomics Comparative Genomics

We are investigating the potential of milk components to influence mammalian development and health, and are using genomics approaches to identify new bioactives for human use. We are looking primarily at bovine milk as a commercial source, but are also using marsupial lactation since it can reveal evolutionally conserved mechanisms in mammals, and because marsupials rely on milk for extended periods of development. For example, the young of the tammar wallaby are born immature after a short gestation period and when born lack an adaptive immune system. We used a bioinformatics approach to identify a number of immune and anti-microbial components in a marsupial EST database derived from genes expressed in the wallaby mammary gland at different stages of lactation. We identified a novel wallaby anti-microbial protein (AGG01) expressed in the early stages of lactation. A direct ortholog to AGG01 is not present in published eutherian genomes, the most closely related genes being others found in the wallaby genome. The gene expression pattern of AGG01 and lysozyme in the mammary gland during lactation suggest that there are two stages of increased immune transfer in the tammar wallaby coinciding with the foetus leaving the womb and the pouch young leaving the pouch. Functional in vitro studies using synthetic forms of AGG01 revealed potent anti-microbial activity against gram-positive bacteria and gram-negative bacteria and a fungus. These may aid the pouch young in adaptation to the environment and increase resistance to potential pathogens. The finding may provide new mechanisms for tackling antibiotic resistant pathogens.

#### FRIDAY 11:15 - 11:30 [F2 CONSERVATION GENETICS 1]

### Genetic variation in koalas on French Island and Kangaroo Island and the likely effect

of contraception protocols on its retention Romane Cristescu<sup>1,\*</sup> Mark Tanaka<sup>1</sup> Cathy Herbert<sup>1</sup> Kris Carlyon<sup>1</sup> Alan Wilton<sup>1</sup> Desley Whisson<sup>2</sup> Kathrine Handasyde<sup>3</sup> Valma Cahill<sup>4</sup> Des Cooper<sup>1</sup>

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Keywords: Conservation Genetics Population Genetics

We have compared microsatellite gene frequencies at 15 loci for French Island in Victoria and the derivative Kangaroo Island population. The number of alleles per locus on French Island has a mean of  $3.8 \pm 0.4$  compared to  $2.4 \pm 0.2$  for Kangaroo Island. The figures for heterozygosity (He) are  $0.48 \pm 0.06$ and 0,41 ±0,05 respectively. Both populations are known to have been through bottlenecks in the recent past, which explains this low genetic variation. As part of the Koala and Kangaroo Contraception Program, we are undertaking a trial of the Suprelorin contraception on koalas. The contraceptive can be administrated in any of three separate protocols. They are (1) regular contraception of females such that all females get at least one opportunity to reproduce, (2) random contraception such that multiple contraception is not avoided, (3) random contraception such that multiple contraception is avoided. We have used population genetics theory to model the effects of these protocols on the retention of genetic variation. The best protocol for retention of genetic variation is (1). Because of the low genetic variability of koalas in southern Australia, consideration of the contraceptive protocol used will be important. Reintroduction of genetic variation through use of males from other populations may become necessary.

WEDNESDAY 15:45 - 16:00 [W4 COMPARATIVE GENOMICS 3]

#### Building a virtual genome using comparative genomics and limited genome sequencing

Brian Paul Dalrymple<sup>1,\*</sup> Representing the International Sheep genomics Consortium <sup>1</sup>CSIRO Livestock Industries

Keywords: Comparative Genomics Bioinformatics

The availability of a complete genome sequence underpins many of the tools used in modern genetics/genomics, such as high-throughput genotyping assays based on SNPs. Despite the availability of many fully, and even more partially, sequenced mammalian genomes, the genome sequence of many species of interest are not high on the sequencing priority list. Here we provide an example of how to combine limited sequence from the organism of interest and the genome sequences of other mammals to create a virtual genome sequence. A high coverage sheep BAC-library was constructed and end-sequenced. By scaffolding the sheep BACs on the current cow, dog and human

genome assemblies around 50% of the ~200 k BACs have been positioned on the human genome with both ends in a tail to tail configuration and between 10 and 500 kb apart. The utilisation of three genomes substantially increased the number of BACs that could be positioned and hence the coverage of the human (and therefore sheep) genome in BAC-comparative genome contigs (BAC-CGCs). Using the sheep marker linkage map the BAC-CGCs were oriented and ordered into our best guess of the structure of the sheep genome. Whilst many breakpoints and rearrangements could be positioned fairly accurately, due to the small number of markers on the sheep map the location and orientation of many of the fragments was based on conserved synteny amongst vertebrate or mammalian genomes. The resulting virtual sheep genome enables the capture of the annotation of the human, dog and cow genomes ordered appropriately for the sheep research community. The virtual genome is an integral part of the development of other sheep genomics research tools, such as a sheep whole genome SNP chip and the planned eventual sequencing of the complete genome.

THURSDAY 11:30 - 11:45 [TH3 FUNCTIONAL GENOMICS 2]

### Investigation of the Immune Strategies for Survival in the Neonatal Tammar (Macropus eugenii).

Kerry Daly<sup>1,\*</sup> Matthew Digby<sup>1</sup> Christophe Lefevre<sup>2</sup> Sonia Mailer<sup>2</sup> Peter Thompson<sup>1</sup> Kevin Nicholas<sup>2</sup> Elizabeth Deane<sup>3</sup> Peter Williamson<sup>1</sup>

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Keywords: Comparative Genomics Functional Genomics

Maruspial young are born in an altricial state, with only those organ system considered essential for perinatal survival well developed. The immune system does not mature until around 90 days in the tammar (Macropus eugenii), which coincides with when the young first leaves the pouch. Prior to this, immune protection of the young is conferred via the secretion of immunoglobulins in the milk of the mother. In this study we examined the expression of immune components in the milk of the mother and in the young themselves. A cDNA library was created from a tammar mammary gland, allowing the identification of the active immune genes. Using cDNA microarrays, which were custom made based on this cDNA library, we analysed the expression of many immune genes in the tammar mammary gland throughout the lactation cycle. Immunoglobulins and components of the innate immune system (such as complement, lysozyme, cathepsins and ferritin) are differentially regulated in the adult tammar mammary gland during periods of increased immune challenge, i.e. birth and leaving the pouch. In the pouch young themselves, we used quantitative PCR to determine gene expression. Pathogen recognition receptors (Toll-like receptors), antimicrobial peptides, non-specific immune cells were detected in all organs examined as early as five days post partum, when at a much earlier stage of development than in their eutherians. Differential expression of immune components in the mother's milk and the earlier activation of the innate immune system, may represent immune strategies that allow the marsupial young to continue development in the pathogen laden environment of the pouch.

FRIDAY 15:30 - 15:45 [F5 ADAPTATION]

### Amino Acid Site-Specific Fitness and Epistasis in HIV-1

Jack da Silva<sup>1,\*</sup>

<sup>1</sup>University of Adelaide

Keywords: Adaptation Evolutionary Genetics Population Genetics

I have recently shown that site-specific amino acid marginal fitness is correlated with amongpopulation mean site-specific amino acid frequency in HIV-1. This provides a simple method of estimating site-specific fitness effects. Here, I explore the fitness effects of interactions among sites, that is, fitness epistasis. Analysis of the intensely studied exterior envelope glycoprotein V3 region reveals strong covariation among sites that may be explained by V3 structure. I show how this covariation is related to fitness epistasis and discuss the application of these methods to investigating the adaptive evolution of HIV-1.

#### TUESDAY 11:00 - 11:15 [T1 COMPARATIVE GENOMICS 1]

### Identifying genes encoding symbiotic transporters in nitrogen-fixing soybeans David Day<sup>1</sup>

<sup>1</sup>University of Sydney

Keywords: Comparative Genomics Comparative

Genomics Symbiotic nitrogen fixation in legumes takes place in specialised structures on the root, nodules, which form when rhizobial bacteria in the soil invade the root. Within the infected cells of the legume root, symbiotic forms of the bacteria, bacteroids, fix atomospheric N2 to ammonia which is

delivered to the plant; in return, the plant provides the bacteroids with carbon and a wide range of other nutrients. The bacteroids within infected cells are surrounded by a membrane to form a facultative organelle called the symbiosome. The host synthesised symbiosome membrane controls nutrient exchange between the partners via a number of transport proteins embedded in the membrane. We have identified a number of these transporters using biochemical assays and are now searching for the genes that encode them. Using a nodule cDNA library and a PCR-aproach based on homology with known gene sequaences in other organisms, we have identified genes encoding transporters for ammonia, malate, zinc and iron. We are in the process of localising the products of these genes and examining their expression profiles and regulation.

#### WEDNESDAY 14:15 - 14:30 [W4 COMPARATIVE GENOMICS 3]

**Mapping genes on tammar wallaby chromosome 5** Janine E Deakin<sup>1,\*</sup> Paul D Waters<sup>1</sup> James Fong<sup>1</sup> Bianca Dobson<sup>1</sup> Cecilia van den Hurk<sup>1</sup> Vidushi Patel<sup>1</sup> Edda Koina<sup>1</sup> Jennifer AM Graves<sup>1</sup>

ARC Centre for Kangaroo Genomics, Research School of Biological Sciences, The Australian National University

Keywords: Comparative Genomics Evolutionary Genetics

Marsupials have proven extremely valuable in comparative genomic studies, as they occupy an important phylogenetic position between birds and eutherian mammals. The tammar wallaby has been the Australian marsupial of choice for genetic studies over the past 30 years. Now the wallaby genome is being sequenced to a depth of 2x coverage and is the first Australian marsupial genome project. To anchor the sequences, we are generating a physical map of genes on wallaby chromosomes. Of particular interest are chromosomes harbouring human X chromosome genes. The mapping of these genes will assist in tracing the evolutionary history of the human X chromosome.

Previous gene mapping studies have shown that wallaby chromosome 5 contains genes from the human X, identifying a region that has been recently added to the X chromosome in eutherians. Chromosome painting has revealed that wallaby chromosome 5 shares homology with opossum chromosomes 4 and 7. By using the opossum genome sequence assembly as a guide, it has been possible to intensely map this region in the wallaby and determine the extent of conserved syntemy between chromosome 5 and the human X. We have found that wallaby chromosome 5 has two blocks of human X genes, separated by genes from human chromosomes 2, 15 and 21. These blocks are found on two chromosomes in the opossum. In addition, we have identified gene blocks corresponding to blocks from human chromosomes 1, 2, 3, 11, 13, and 19 and detected rearrangements between opossum and wallaby that remain undetectable in chromosome painting experiments. Our data highlights the need for comparative gene mapping and the important role the tammar wallaby plays in comparative genomics studies.

THURSDAY 11:30 - 11:45 [TH2 EVOLUTIONARY GENETICS]

Mechanisms of gene transfer from the chloroplast to the nucleus

Sven K. Delaney<sup>1,\*</sup> Gregor W. Schmidt<sup>1,2</sup> Anna Sheppard<sup>1</sup> Jeremy N. Timmis<sup>1</sup>

<sup>1</sup>University of Adelaide

<sup>2</sup>Friedrich Schiller University of Jena

Keywords: Evolutionary Genetics Plant Genetics

DNA transfer from the chloroplast and mitochondrial genomes to the nuclear genome has occurred extensively during evolution of the eukaryotic cell. Transferred sequences will be expressed only if they acquire appropriate nuclear regulatory sequences, suggesting that functional DNA transfer is a rare event. However, in tobacco (Nicotiana tabacum) functional genes are transferred from the chloroplast to the nuclear genome at a rate of approximately 1 nuclear integration per 16,000 pollen grains (Huang et al., 2003; Nature 422: 72-76), and the rate of non-functional gene transfer may be considerably higher. This has important implications for the containment of transgenes in the chloroplast genome, and suggests the existence of mechanisms that promote the bulk transfer of DNA from the chloroplast to the nucleus.

We have investigated these mechanisms in the context of both germ line and somatic cells. Chloroplasts in the developing tobacco pollen are progressively degraded during microsporogenesis, but analysis of gene transfer by quantitative RT-PCR did not reveal significant chloroplast to nucleus DNA transfer. This result suggests that chloroplast to nucleus DNA transfer may occur at other points during development, and is being confirmed in ongoing experiments. In leaf and other somatic cells, high frequency spontaneous transfer of a functional marker gene from the chloroplast to the nucleus was observed. The effects of common abiotic stresses on the frequency of this transfer were also determined.

#### FRIDAY 14:00 - 14:15 [F4 CONSERVATION GENETICS 2] Dramatic Variations in Microbial Communities among Coral Reef Ecosystems Elizabeth A Dinsdale<sup>1</sup> <sup>1</sup>San Diego State University Keywords: Environmental Microbes

Microbes are implicated in coral reef decline, but specific mechanisms causing decline remain unknown. Metagenomic, microscopic and culturing techniques applied on coral reefs across four atolls in the central Pacific showed increases in abundance of virus-like particles, Bacteria, Archaea, and protists in the overlying waters. Taxonomic and metabolic gene composition of eight metagenomes, four that captured the Bacteria and Archaea and four that captured the viral fraction of the coral reef water microbial community were assessed. Analysis of the Bacteria and Archaea using three databases demonstrated a shift from a balance of autotrophy and heterotrophy to a predominantly heterotrophic community. Moreover, on the atoll that had the highest prevalence of coral disease, many heterotrophic Bacteria showed sequence similarities to potential pathogens. The phages, identified from the viral metagenomes, were classified as infecting either autotrophic or heterotrophic hosts and showed a similar switch as that described in the bacterial and archaeal community across the four atolls. The relative proportions of the functional genes on each atoll confirmed the non-linear change in the microbial community. On the atoll with no people, the proportion of genes associated with photosynthesis was 3.4 % and this increased to 44.3 % on the atoll with a moderate number of people. On the atoll with the largest number of people, the photosynthetic genes represented only 0.3 % of the metagenome, whereas genes associated with carbon utilization comprised 24 %. The changes in the microbial community were dramatic and correlated with numbers of people on each atoll, suggesting that anthropogenic effects on microbial communities contribute to coral decline.

#### WEDNESDAY 11:15 - 11:30 [W1 COMPARATIVE GENOMICS 2]

#### Meta-analysis of metagenomes: metabolic fingerprints of microbial and viral communities from eight environments.

Elizabeth A Dinsdale<sup>1,\*</sup> Dana Hall1 Forest Rohwer1 Robert A Edwards1 <sup>1</sup>San Diego State University Keywords: Functional Genomics

Microbial and viral communities are vital for processing energy and nutrients through the ecosystem. Understanding the processes and metabolism of each ecosystem is central to ensuring the health and balance of the environment. The metabolic potential of 87 metagenomes sequenced from eight major environs, including subterranean, salt lakes, marine and freshwater locations and within organism such as, fish, corals, microbialites and terrestrial animals, was analyzed by comparing more than 1.25 million sequences to the known databases. Distinct environmental metabolic fingerprints were identified for both microbial and viral communities. Several metabolic processes such as respiration, protein metabolism, sulfur metabolism, motility and chemotaxis, structured much of the difference between communities. Several of the important metabolic processes within the microbial community were mirrored within the viral community, providing further insight into the interactions between these two assemblages.

#### THURSDAY 12:15 - 12:30 [TH1 POPULATION GENETICS 1]

Population structure of the Giant Australian Cuttlefish - implications for the world's **largest breeding aggregation of cephalopods** Steve Donnellan<sup>1,\*</sup> Bronwyn Gillanders<sup>2</sup> Melita de Vries<sup>2</sup>

South Australian Museum

<sup>2</sup>University of Adelaide

Keywords: Population Genetics Speciation and Phylogeography

The Giant Australian Cuttlefish (Sepia apama) is unique among cephalopods for its annual mass breeding aggregation in the upper Spencer Gulf, South Australia. Recent changes in fishing practice and the imminent construction of a desalination plant could threaten the mass breeding population. We have investigated population structure, using microsatellites, mitochondrial DNA, morphology and statolith microchemistry, to determine the geographic extent of the population that breeds in the upper Spencer Gulf. All of the data sets are concordant in showing the upper Spencer Gulf population is distinct from the rest of the species range in South Australia. We discuss the systematic and conservation implications of these findings and evolutionary and oceanographic explanations for the pattern.

#### WEDNESDAY 11:30 - 11:45 [W1 COMPARATIVE GENOMICS 2]

### The evolution of a novel mitochondrial genome structure in the cyst-forming nematodes

Mark Dowton<sup>1,\*</sup> Tracey Gibson<sup>1</sup> Angelique Riepsamen<sup>1</sup> Vivan Blok<sup>2</sup> Mark Phillips<sup>2</sup> Ian Riley<sup>3</sup> <sup>1</sup>School Biology, University of Wollongong <sup>2</sup>Scottish Crop Research Institute <sup>3</sup>SARDI

Keywords: Evolutionary Genetics Comparative Genomics

We sequenced four circular, mitochondrial subgenomes from the potato cyst nematode Globodera pallida. These subgenomes contained overlapping subsets of the 37 genes normally found in animal mitochondrial genomes, a feature not previously reported for any other animal species. A previous search for full length genomes within this nematode failed to find any. Analysis of the sequence data indicate that three of these subgenomic mitochondrial circles are mosaics, comprising long, multigenic fragments derived from fragments of the other circles. This pattern is consistent with the operation of intermitochondrial recombination, a process generally considered absent in animal mitochondria. Many of the duplicated genes contain deleterious mutations (primarily point indels that disrupt the reading frame), but sequencing of multiple copies of these genes indicates that there is variation at polythymidine tracts. Comparison of the genomic sequences with cDNA is consistent with the operation of insertion/deletion editing of mitochondrial transcripts.

We then sequenced five mitochondrial genomes from the close relative, Globodera rostochiensis, and found that these were similarly subgenomes, each containing only a subset of the 37 genes normally found on animal mitochondrial genomes. Variation at polythymidine tracts was also evident, and comparison of genomic sequences with an EST database of this nematode similarly was consistent with the operation of insertion/deletion editing. We are currently investigating the mitochondrial structure of the heteroderid nematodes, in order to more accurately determine the evolutionary origin of these novel mitochondrial structures.

#### TUESDAY 11:45 - 12:00 [T1 COMPARATIVE GENOMICS 1]

#### Sex microchromosomes in Australian dragon lizards

Tariq Ezaz<sup>1,\*</sup> Alex Quinn<sup>2</sup> Stephen Sarre<sup>2</sup> Arthur Georges<sup>2</sup> Jenny Graves<sup>1</sup>

<sup>1</sup>Comparative Genomics Group, Research School of Biological Sciences, The Australian National University, ACT 0200, Australia

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Keywords: Comparative Genomics Evolutionary Genetics

Reptiles exhibit an impressive array of sex-determining modes, including GSD (genetic sex determination) with male (XX/XY) and female (ZZ/ZW) heterogamety in turtles and lizards, and GSD with female heterogamety in snakes. Many lizards and turtles have GSD with cryptic sex chromosomes (no obvious heteromorphy) while many other lizards and turtles, and all crocodilians, have TSD (temperature-dependent sex determination). Dragons lizards are particularly interesting, because their evolutionary history suggests many recent transitions between GSD and TSD mechanisms.

Unlike mammals, reptilian and avian genomes are composed of two distinct types of chromosomes: macro and micro. In many lizards, including some Australian agamids, the sex chromosomes are a microchromosome pair, often cryptic and one member of the pair may be highly heterochromatic. Are the sex microchromosomes of Australian agamids evolutionarily conserved or have they evolved independently in different species? Are they evolutionarily advanced or primarily junk DNA? Are sex microchromosomes present in TSD agamids? Do they play any role in transitions between GSD and TSD?

To explore these questions, we applied comparative genomic hybridization to identify and characterise cryptic sex microchromosomes in several species of Australian dragons. We isolated a short sequence of sex chromosomal DNA from the Central bearded dragon (Pogona vitticeps), and expanded this into a hybridisation probe by genome walking. This probe was used for chromosome painting of other Australian dragons (both TSD and GSD species) to investigate the relationship of sex microchromosomes in this group.

#### FRIDAY 15:00 - 15:15 [F4 CONSERVATION GENETICS 2]

## Phylogeography of an endangered freshwater fish, Macquarie perch (Macquaria australasica) in Eastern Australia.

Leanne K Faulks<sup>1,\*</sup> Dean M Gilligan<sup>2</sup> Luciano B Beheregaray<sup>1</sup> <sup>1</sup>Molecular Ecology Lab Macquarie University, <sup>2</sup>New South Wales Department of Primary Industries

Keywords: Speciation and Phylogeography Conservation Genetics

Macquaria is a freshwater fish genus of substantial importance to Australian fisheries management. One member of the genus, Macquaria australasica, has a restricted distribution in the upper reaches of streams in eastern Australia (Murray-Darling, and some south eastern NSW coastal basins). Habitat degradation, competition, and disease, are threatening the persistence of M. australasica which is now classified as endangered in NSW. Here we investigate historical patterns of population genetic structure and diversification in M. australasica. A comprehensive collection of samples has been obtained from across the species range and mitochondrial DNA control region sequence data were used to reconstruct the genealogical history of populations based on coalescence methods. Macquarie perch displays marked phylogeographic structure with divergent populations in the Murray- Darling and coastal basins of south eastern NSW. The identification of distinct lineages will assist in the conservation and recovery planning for the species and give insight into the evolution of drainage basins in eastern Australia. The results presented here form part of a larger project on the Macquaria genus expected to provide a valuable contribution to the fields of comparative phylogeography, conservation genetics, and fisheries management in Australia.

WEDNESDAY 15:00 - 15:15 [W6 FUNCTIONAL GENOMICS 1]

Adaption in the Hawaiian Picture-winged Drosophila Marla A Fisher<sup>1,\*</sup>

<sup>1</sup>Department of Biology, University of Hawaii, Hilo, HI, USA

**Keywords:** Adaptation Speciation and Phylogeography Comparative Genomics Functional Genomics Population Genetics

The Hawaiian Picture-winged *Drosophila* represent approximately 120 of 800-1000 species of *Drosophila* that are endemic to Hawaii. These Picture-winged *Drosophila* are considered an excellent example of adaptive radiation on the Hawaiian Islands. Yet, little is known about evolved adaptive changes that may underlie local adaptation and perhaps incipient speciation in these species. We propose to use Agilent oligonucleotide arrays to screen for gene expression changes that may be indicative of local adaptation. *Drosophila crucigera* occurs in different habitats (wet versus dry slopes) on the island of Oahu, and also on the island of Kauai. Thus, it is a good species to examine genes that may be involved in local adaptation and/or incipient speciation among habitats. We expect that we will observe similar genes that may differ across locations and between islands. We will use the sequenced genome of the Picture-winged *Drosophila grimshawi* for the microarrays. We will also sample loci across the *D. crucigera* genome in order to obtain an understanding of effective population size and genetic distance, and to distinguish genes that may differ due to drift from that due to selection.

#### WEDNESDAY 12:30 - 12:45 [W3 BIOINFORMATICS]

**Modelling the Omics Network of Hepatocellular Carcinoma Using Visual Graph Mining** David Cho Yau Fung<sup>1,\*</sup> John Galloway<sup>2,3</sup>

<sup>1</sup>Independent Investigation, South Strathfield, NSW 2136, <sup>2</sup>NetMap Analytics Pty. Ltd., St Leonards, NSW 2065, <sup>3</sup>Complex Systems Research Centre, University of Technology Sydney, NSW 2001 **Keywords:** Bioinformatics Functional Genomics

Modularity is an important topological feature of biological networks such as a proteome network, a gene co-expression network, signal transduction pathways, and metabolic pathways. In essence, modules can be defined as sub-networks whose entities are more frequently connected to each other than entities outside the sub-networks. When visualized as a node-edge graph, modules represent communities of high connectivity. Because it exists in a variety of biological context, modularity should be seen not only in physical networks but also integrated networks of physical and ontological entities. The authors called the latter type of networks as the 'omics' network. Visual graph mining can be used to compare the 'omics' networks across clinical phenotypes that may reveal the molecular pathology of hepatocellular carcinoma. In particular, the topological change of certain modules due to the change of membership within should reveal the molecular relationships that may explain the progression of the disease. In this paper, the authors demonstrated the use of visual graph mining in modeling the omics network of hepatocellular carcinoma. Keywords: visual graph mining, modularity, omics network, hepatocellular carcinoma.

#### WEDNESDAY 12:00 - 12:15 [W3 BIOINFORMATICS]

### Gentrepid: A Bioinformatics System For Candidate Disease Gene

Richard A George<sup>1,2</sup> Jason Y Liu<sup>1</sup> Robert J Bryson-Richardson<sup>2</sup> Diane Fatkin<sup>2,3</sup> Merridee A Wouters<sup>1,3,\*</sup>

<sup>1</sup>Structural & Computational Biology, VCCRI, <sup>2</sup>VCCRI, <sup>3</sup>School of Medicine, UNSW Keywords: Bioinformatics Gene and QTL Mapping

The identification of genes responsible for human disease enables an understanding of disease mechanism and is essential for the development of diagnostics and therapeutics. Linkage analysis of disease inheritance patterns is a successful procedure to associate a disease with a specific genomic region. Unfortunately, isolating the disease-causing gene(s) can be difficult: genomic regions are often large, containing hundreds of candidate genes, making experimental methods time-consuming and expensive. Furthermore, searches for single nucleotide polymorphisms (SNPs) in the genomes of individual patients from clinical studies will produce a large number of potential gene candidates. Clearly, these high-throughput analyses will require computational approaches to identify good candidates for further study.

We present two computational approaches to prioritize candidate genes for further experimental study: Common Module Profiling (CMP) and Common Pathway Scanning (CPS). CMP is based on the hypothesis that disruption of genes of similar function will lead to the same phenotype and identifies likely candidates using a domain-dependent sequence similarity approach. CPS is based on the assumption that common phenotypes are associated with dysfunction in proteins that participate in the same complex or pathway and applies network data derived from protein-protein interaction and pathway databases to identify relationships between genes. Both algorithms use two forms of input data: known disease genes or multiple disease loci. When using known disease genes as input, our combined methods have a sensitivity of 0.518 and a specificity of 0.966 and reduce the candidate list by 13-fold. When using multiple loci, our methods successfully identify disease genes for all benchmark diseases with a sensitivity of 0.835 and a specificity of 0.626. Our combined approach prioritizes good candidates and will accelerate the disease gene discovery process.

#### FRIDAY 14:45 - 15:00 [F4 CONSERVATION GENETICS 2]

### Capturing maximum genetic diversity in a minimum sample size using molecular markers and ecogeographical data. Kioumars Ghamkhar<sup>1,\*</sup> Richard Snowball<sup>2</sup>

<sup>1</sup>Centre for Legumes in Mediterranean Agriculture, University of Western Australia <sup>2</sup>Australian Trifolium Genetic Resources Centre, Department of Agriculture and Food WA

Keywords: Evolutionary Genetics Phylogeny, Biodiversity & Barcoding Plant Genetics **Bioinformatics Conservation Genetics** 

Although molecular techniques have been used to assess the genetic diversity of some species of Trifolium L. or within the genus generally, there is no record of using these techniques to develop core collections within the genus or evaluate genetic diversity of T. spumosum L. Our study aimed to develop a two step approach for choosing a core collection of T. spumosum as a model approach for other pasture legumes. First, a total of 317 accessions, with near-complete ecogeographical data, were selected from the Australian ex situ collection of T. spumosum. Missing data, including details of geographic ditribution were completed. A preliminary core of about 30% of the collection was, then, selected.

Fluorescent AFLP (fAFLP) was applied for molecular screening of genetic diversity in the collection's first subset. Four of the most ecogeographically diverse accessions were screened in search of the primers with the highest number of bands and polymorphism. By combining the results of this screening and ecogeographical data we developed the final core collection of 32 accessions, which contains 30% of the first subset.

Our study demonstrated that a combination of AFLP markers and ecogeographical data can be used to develop an effective core collection that maintains most of the genetic diversity of and represents the original collection. Such a core will allow breeders to more effectively select cohorts for field testing and enable gene bank managers to more efficiently conserve germplasm. It will also help identify gaps in genetic diversity, resulting in a more effectively targeted collection in future. The quality of ecogeographical data of collection sites is critical to the success of this approach to developing a core collection. This model can be expanded to sampling accessions for phylogenetic studies of other crops and native plants with large collections.

#### FRIDAY 14:00 - 14:15 [F5 ADAPTATION] Improving the mass-rearing of Queensland Fruit Fly Stuart Gilchrist<sup>1,\*</sup> <sup>1</sup>Fruit Fly Research Centre, University of Sydney

Keywords: Adaptation Population Genetics

As pesticide use is being continually curtailed, biologically benign control methods like the sterile insect technique (SIT) will become increasingly important. A pre-requisite for SIT is the ability to rear the pest in large numbers. One such insect factory, rearing the Queensland fruit fly, Bactrocera tryoni, has been operating near Sydney since 1998. It can produce over 10 million flies per week. While numerous quality control issues associated with mass-rearing have been already addressed, the genetics of this mass-rearing effort has received little attention. Life history theory suggests that selection for the desirable qualities of a mass-reared strain (laboratory adaptation and high fecundity) are likely to reduce traits important for field performance (dispersal ability, longevity and stress resistance). This presentation will consider the changes that have occurred during domestication and the methods by which an optimal balance between factory and field performance might be achieved in practice. The results are relevant not only to the improved control of Queensland fruit fly. Even more damaging Bactrocera species have already, and will in the future, invade Australia. The present work serves as a blueprint for future SIT campaigns against exotic fruit fly incursions.

#### WEDNESDAY 11:00 - 11:15 [W3 BIOINFORMATICS]

### Comparison of multiple sequence alignment methods, with implications for surveys of proteomic data

Tanya Golubchik<sup>1,\*</sup> Michael J Wise<sup>2</sup> Simon Easteal<sup>3</sup> Lars S Jermiin<sup>1</sup>

<sup>1</sup>School of Biological Sciences, University of Sydney

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<sup>3</sup>John Curtin School of Medical Research, Australian National University

Keywords: Bioinformatics Comparative Genomics Functional Genomics Phylogeny, Biodiversity & Barcoding

Aligning several sequences of nucleotides or amino acids to construct a multiple sequence alignment (MSA) is a crucial first step in the analysis of genomic and proteomic data. In particular, alignment of highly similar sequences is important for the analysis of protein data, where many such sequences would be expected as a result of alternative splicing. We assessed the performance of six popular MSA programs (CLUSTALW, DIALIGN-T, MAFFT, MUSCLE, PROBCONS and T-COFFEE), and one experimental program, PRANK, on protein sequences that differed only by the placement of gaps. On datasets containing overlapping gaps, most MSA programs preferentially aligned the gaps vertically, even at the expense of incorrectly aligning homologous residues in the flanking regions. Of the programs assessed, only DIALIGN-T was able to place overlapping gaps correctly relative to one another, but this was context-dependent, and was observed only on some of the datasets. On alignments with non-overlapping gaps, both DIALIGN-T and MAFFT (G-INS-I) were able to align gaps with near-perfect accuracy, but only MAFFT produced the correct alignment consistently. When applied to alignments of multiple isoforms of alternatively spliced genes, the same was true: both DIALIGN-T and MAFFT produced highly accurate alignments, with MAFFT being the more consistent of the two. Other programs, notably T-COFFEE and CLUSTALW, were less accurate. For all datasets, alignments produced by different MSA programs were significantly different, indicating that reliance on a single MSA program may give misleading results. It is therefore advisable to use more than one MSA program when dealing with such sequences, particularly for high-throughput and pipeline applications where manual refinement of each alignment is not practicable.

#### WEDNESDAY 14:45 - 15:00 [W5 PHYLOGENY]

**Genetic divisions of the Collared peccary from the Americas** Jaime Gongora<sup>1,\*</sup> Chris Moran<sup>1</sup>

<sup>1</sup>University of Sydney

Keywords: Phylogeny, Biodiversity & Barcoding Conservation Genetics

The Collared peccary (Pecari tajacu) is one of the three extant recognised species of the family Tayassuidae living in the Americas. To understand phylogenetic relationships among Collared peccaries, two mitochondrial and four nuclear DNA markers of specimens from six countries were analysed. Phylogenetic analyses of mitochondrial sequences showed that Collared peccaries clustered in two major clades, representing North-Central American and South American specimens. Collared peccaries from Colombia split between these major clades. Analyses of nuclear sequences showed a distribution of DNA variants which was consistent with mitochondrial sequence analyses. However,

there was an uncoupling of nuclear and mitochondrial sequence variation in two specimens from Colombia. A comparison of pairwise genetic distances between mitochondrial sequences showed that the geographically widespread Collared peccary consists of two unique lineages as genetically distinct as the White-lipped and Chacoan peccaries as well as recognised species of the related Suidae family. This study has implications for breeding programs and efforts to conserve this species. It has also provided a phylogenetic framework to assess captive specimens of unknown origin in Australia and to assess claims to the 'new' Giant peccary species in the Amazonian forest in Brazil.

WEDNESDAY 12:00 - 12:15 [W1 COMPARATIVE GENOMICS 2]

#### Evolution of Cytochrome P450 Structures

Bagavananthem Andavan Gowri Shankar<sup>1,\*</sup> Michael Charleston1 Michael Murray<sup>2</sup> David Hibbs<sup>2</sup> <sup>1</sup>School of Information Technologies, University of Sydney <sup>2</sup>Faculty of Pharmacy, University of Sydney Keywords: Comparative Genomics Bioinformatics

Cytochrome oxidase P450 (CYP) is one of the most important enzymes in metabolizing drugs in humans and other species. These enzymes are present in all species, from archaea and bacteria to fungi and mammals. This study aims at understanding the evolution of this enzyme: How the structures have changed over time and between species and the significant changes in the drug binding site, which alters the function of the protein. From an evolutionary analysis of 15 CYP protein structures, we found that the structure has changed considerably. From the evolutionary trees for these enzymes it is evident that there is a correlation between the structural differences and evolutionary distances between species. Looking at the binding sites of these enzymes we were able to find that there is a pattern in these sites that is conserved over two billion years. In archaea there is a slight change in the pattern which makes it different from the other species. From further consideration of the CYP proteins, we note that the structure in archaea is more complex than in the much more recently diverged humans. There is a histidine to argentine change in the binding site of mammals from archaea and bacteria which plays a major role in determination of the specificity of the protein to drugs. Our work is unique in that it incorporates evolutionary changes with the protein structural and the binding site changes. From earlier phylogenetic analyses, it has been concluded that the protein structures have evolved with very few changes in the binding sites and perform the same function. In our analysis it is evident that there have been substantial structural changes in the structure among species and less change in the binding sites, but the function of the proteins changes with time.

#### TUESDAY 12:00 - 12:15 [T1 COMPARATIVE GENOMICS 1]

#### Genome organisation in platypus and chicken sperm

Frank Grützner<sup>1,\*</sup> Natasha Dodge<sup>1</sup> Julia Mohr<sup>1</sup> Colin K Kremitzki<sup>2</sup> Kyriena Schatzkammer<sup>2</sup> Tina Graves<sup>2</sup> Wesley Warren<sup>2</sup> Enkhjargal Tsend-Ayush<sup>1</sup>

<sup>1</sup>School of Molecular & Biomedical Science, The University of Adelaide, 5005 SA, Australia

<sup>2</sup>Genome Sequencing Center, Washington University School of Medicine, St Louis, MO 63108, USA **Keywords:** Comparative Genomics Evolutionary Genetics

In mammals non-random positioning of chromosomes within the sperm nucleus is a well-established fact. Moreover, for the mammalian X evolutionary conservation of the localisation has been shown in different species. In contrast the chicken genome, which is divided into large genepoor macrochromosomes (MACs) and small gene-rich microchromosomes (MICs), hybridisation of repetitive elements showed that chromosomes are randomly distributed in the chicken sperm, although a preferentially central localisation of MICs was observed. To get a more accurate picture of genome organisation in chicken sperm, we mapped chromosome specific BAC clones of chicken MICs and sex chromosomes on chicken sperm. Our preliminary results indicate that in contrary to previously published results some MICs occupy a more peripheral position. In addition we obtained first results indicating that not all MICs are randomly organised in chicken sperm.

The unique evolutionary position of monotremes gives valuable insight into the evolution of genome organisation in mammals, in particular in comparison with the entirely different genome organisation in chicken. The elongated monotreme sperm provides an ideal system to test current ideas about genome organisation in sperm. The platypus karyotype contains chromosomes similar sized as MICs as well as large chromosomes. In addition they feature an extraordinary ten sex chromosome system, which shows homology to chicken sex chromosomes. In platypus we selected BAC clones representing autosomes of different size, platypus chromosome 6, which shares homology with the human X chromosome and sex chromosome specific BAC clones. We found a non-random distribution of chromosomes in platypus sperm. In addition platypus chromosome 6 did not localise to the anterior as expected from its homology to the X chromosome of other mammals. Interestingly

some of the Y chromosomes cluster at the posterior part of the sperm, which might reflect organisation of sex chromosomes beyond the first meiotic division.

#### THURSDAY 12:00 - 12:15 [TH2 EVOLUTIONARY GENETICS]

Sex linked genetic influence on caste determination in a termite

Yoshinobu Hayashi1 Hitoshi Miyata1 Nathan Lo<sup>2,\*</sup> Osamu Kitade<sup>1</sup>

<sup>1</sup>Natural History Laboratory, College of Science, Ibaraki University, Mito, Ibaraki 310-8512 Japan <sup>2</sup>Behaviour and Genetics of Social Insects Laboratory, School of Biological Sciences, The University of Sydney, NSW, Australia 2006

Keywords: Evolutionary Genetics

The eusocial insects termites are renowned for their extreme morphological specialization between castes, their large and highly populated mound structures, and their dominant role in tropical ecosystems throughout the world. The bulk of species associated with these characteristics have both a nymph caste (that develops into the imaginal form) and an irreversibly wingless worker caste (that almost always remains sterile). The early developmental bifurcation separating these castes is widely accepted to be strictly environmentally determined. We have found the first evidence for a genotypic influence over this process. Crossing of worker and nymph derived neotenic reproductives resulted in highly differentiated offspring caste and sex ratios, despite uniform rearing conditions. The data fit an X-linked, one-locus model, with unusual properties. The proposed genetic influence can be overridden under normal conditions by environmental factors, providing the colony with flexibility over worker production.

#### FRIDAY 12:00 - 12:15 [F1 POPULATION GENETICS 2]

A population genetic study of one of Australia's most invasive fish species, the **common carp (Cyprinus carpio), in the Murray-Darling River System** Gwilym David Haynes<sup>1,\*</sup> Frank Nicholas<sup>1</sup> Dean Gilligan<sup>2</sup> Peter Grewe<sup>3</sup>

<sup>1</sup>University of Sydney, Invasive Animals CRC

<sup>2</sup>Department of Primary Industries, Invasive Animals CRC

<sup>3</sup>CSIRO, Invasive Animals CRC

Keywords: Conservation Genetics Population Genetics

Introduced to Australia in the late nineteenth century, common carp have become a major freshwater pest. Thriving in disturbed habitats, they degrade waterways and compete with native fish for resources. Their mobile nature and fecundity, along with illegal human assisted dispersal, has allowed them to colonise nearly all of the Murray-Darling river system, as well as many coastal rivers. Their broad tolerance to salinity and temperature gives them the potential to occupy possibly all permanent freshwater habitats in Australia. Genetic factors are likely to have played a key role in the success of common carp in Australia. Common carp were present in low densities in the Murray-Darling River System as early as the 1920s, but did not become highly invasive until the 1960s, when a new strain of carp was illegally introduced from Germany.

This project explores the population genetics of common carp with the aim of making recommendations for control programs. Preliminary results so far indicate that there have been multiple introductions of carp into Australia at different times; that a large, panmictic carp population is present in the low-lying parts of the Murray-Darling Basin; and that the dams surrounding these low-lying regions act to limit dispersal. Information such as this, when combined with demographic data and population modelling, will be used in the management of this exotic pest.

#### **TUESDAY 19:25 - 19:40 [S1 STUDENT]**

Insight into the limits of the Japanese encephalitis virus through a better understanding of the biodiversity of its mosquito vector. Stéphane Hemmerter<sup>1,\*</sup> Jan !lapeta<sup>2</sup> Nigel W. Beebe<sup>1</sup>

Institute for the Biotechnology of Infectious Diseases, University of Technology

<sup>2</sup>Faculty of Veterinary Science, University of Sydney

Keywords: Phylogeny, Biodiversity & Barcoding Speciation and Phylogeography

The mosquito Culex annulirostris Skuse (Diptera: Culicidae) is the major vector of endemic arboviruses in Australia and is also responsible for the establishment of the Japanese encephalitis virus (JEV) in southern Papua New Guinea (PNG) as well as its incursions into northern Australia. Regular JEV activity occurs on the islands separating PNG and Australia, yet JEV has not established on mainland Australia despite the abundance of Cx. annulirostris and porcine amplifying hosts. We

address the hypothesis that detailing the genetic diversity in *Cx. annulirostris* will shed light on the paradox that JEV can cycle just 70 km north of Australia yet not establish on mainland Australia.

We sequenced 538 bp of the mitochondrial DNA cytochrome oxidase I gene from 273 individuals collected from 43 localities in Australia and the southwest Pacific region to describe the phylogeography of *Cx. annulirostris*. Maximum Likelihood and Bayesian analyses reveal supporting evidence for multiple divergent lineages that display geographic restriction. *Culex annulirostris* contained five geographically restricted divergent lineages, with one lineage restricted to the Solomon Islands and two identified mainly within Australia. The other two lineages were restricted to PNG and the Torres Strait Islands with a southern limit at the top of Australia's Cape York Peninsula.

These results help explain the difficulty of adult morphology identification and the ecological diversity of *Cx. annulirostris*. Notably, the southern limit of the two PNG-lineages of *Cx. annulirostris* coincides exactly to the current southern limit of JEV activity in Australasia and we suggest that biological variation in these COI lineages may explain why JEV has not established yet on mainland Australia. This information is also valuable in studying mosquito-borne disease in Australia and for the rational design of JEV vector competence experiments.

#### WEDNESDAY 12:00 - 12:15 [W2 PHYLOGEOGRAPHY]

#### Comparative phylogeography of two long-necked turtle species in the Murray-Darling Basin demonstrate how molecular data can answer ecological questions Kate Hodges<sup>1,2,\*</sup>

<sup>1</sup>University of Adelaide, Environmental Biology

<sup>2</sup>University of Canberra, Institute for Applied Ecology

Keywords: Speciation and Phylogeography Phylogeny, Biodiversity & Barcoding

The broad-shelled turtle Chelodina expansa, and the common longneck turtle Chelodina longicollis are co-distributed throughout the Murray-Darling Basin (MDB) and in some east coast river systems. Using a 600bp fragment of the mitochondrial ND4 gene, comparisons of phylogeographic structure between the two species revealed historical barriers to gene flow and possible colonisation routes for the MDB, which during the ice-ages is likely to have been largely unsuitable for these turtles. The broad-shelled turtle has a very shallow gene tree and very little diversity within the MDB, while the common longneck has two major mtDNA lineages each with little diversity within the MDB. Moreover in the MDB both species have substantially less diversity compared with coastal rivers of eastern Qld and NSW. This is indicative of recent and rapid population expansions in the MDB. Comparisons of phylogeographic structure in both species suggests that the sources of these expansions are likely catchments in north eastern New South Wales and south east Queensland. Results drawn from this comparative approach also demonstrate the power of molecular markers in addressing ecological questions. Throughout its temperate zone range the broad-shelled turtle characteristically nests in autumn and winter, a pattern more typical of warm-temperate and tropical turtles. If the broadshell turtle population in the MDB was originally founded by populations in south east Queensland, such an expansion from ancestral sub-tropical waters into the cool-temperate MDB could explain why this species exhibits such an unusual nesting ecology. The expansion may have been characterised by a strong founder effect, which could have limited genetic variation in nesting behaviour and physiology and thus the potential to evolve spring nesting which is more typical of cool-temperate climate turtles.

#### FRIDAY 14:15 - 14:30 [F5 ADAPTATION]

**Linking field fitness with genetic polymorphisms under thermal selection in Drosophila** Ary Hoffmann<sup>1,\*</sup> Steve McKechnie<sup>2</sup> Lea Rako1 Mark Blacket1

<sup>1</sup>The University of Melbourne

<sup>2</sup>Monash University

Keywords: Adaptation Evolutionary Genetics

Lists of candidate genes for thermal adaptation are emerging from studies of comparative genomics, microarray comparisons, and quantitative genetic analyses. However so far there have been few cases that successfully link genetic polymorphisms in these genes to adaptive shifts in natural populations. We are testing the feasibility of using a combination of clinal studies, field releases, field cage experiments and laboratory tests to develop such links in Drosophila melanogaster from the tropical-temperate cline in eastern Australia. We illustrate the approach with recent data collected on the frost, Dca, Adh and hsp68 polymorphisms.

#### TUESDAY 18:25 - 18:40 [S1 STUDENT]

## Testing estimates of genetic population structure: levels of confidence in population genetics

Clare E Holleley<sup>1,\*</sup> William B Sherwin1 Richard A Nichols<sup>2</sup> <sup>1</sup>The University of New South Wales <sup>2</sup>Queen Mary, University of London **Keywords:** Population Genetics Conservation Genetics

Habitat fragmentation is recognized as a major contributing factor to the risk of population extinction in the wild. A consequence of habitat fragmentation is that genetic exchange (or gene flow) is limited between populations. Limited gene flow (Nem) may promote genetic differentiation among subpopulations, resulting in genetic population structure. As the effects of habitat fragmentation can be detected via genetic indicators, current conservation efforts often rely on estimating population structure in wild populations. Whilst there is a wide range of genetic methods to estimate population structure, none have been formally tested in live organisms. In controlled replicated Drosophila melanogaster experiments, we have empirically tested three current methods of estimating genetic population structure: FST, RST and phi via analysis of molecular variance (AMOVA). Using eight microsatellites we calculated the equilibrium value of FST, RST and phi in the experimental populations of known effective population size (Ne = 14.15) and dispersal rate (m = 0.0025-0.04). We then compared the empirically determined values to those predicted via the conversion of population structure to gene flow. The experimental populations all adhered to Wright's demographic island model, but displayed significantly lower estimates of FST, RST and phi than the values expected under that model. These results strongly suggest that even in the best-case scenario, current methods have the potential to significantly underestimate population structure when applied to real populations. This is of great concern to conservation efforts that rely on genetic methods to detect the effects of habitat fragmentation. These results may call into question the confidence that biologists may have in some of the most widely used molecular tools in conservation biology, evolutionary and population genetics.

WEDNESDAY 15:00 - 15:15 [W4 COMPARATIVE GENOMICS 3] The evolution of genes regulating genomic imprinting in mammals. Timothy A Hore<sup>1,\*</sup> Janine E Deakin1 Jennifer A Marshall Graves1 <sup>1</sup>The Australian National University, Canberra

Keywords: Epigenetics Comparative Genomics Evolutionary Genetics

For the large majority of genes in diploid organisms, alleles inherited from both parents are expressed equally. However, genes subject to genomic imprinting show preferential expression of one allele, favouring either the copy derived from the mother or the copy from the father. How these alleles "remember" which parent they were inherited from is currently not known, although it is thought that parents produce sex-specific epigenetic marks during gametogenesis. These initial marks (or imprints) are later interpreted in offspring, ultimately causing parent-of-origin specific gene expression. In humans and mice, it has been proposed that two closely-related genes, BORIS (Brother Of Regulator of Imprinted Sites) and CTCF (CCCTC-Binding Factor) are involved in the establishment and interpretation of parental imprints for at least one major imprinted loci [1,2]. We examined the evolution of these genes upon the arrival of genomic imprinting in mammals, by studying their orthologous counterparts in the oldest mammalian clades; marsupials (which posses imprinting) and monotremes (thought to predate imprinting). We found that CTCF and BORIS are functionally different in these mammalian groups compared to humans and mice, impacting on current theories regarding their role in imprinted gene regulation.

1. Loukinov DI, Pugacheva E, Vatolin S, Pack SD, Moon H, et al. (2002) BORIS, a novel male germ-line-specific protein associated with epigenetic reprogramming events, shares the same 11-zinc-finger domain with CTCF, the insulator protein involved in reading imprinting marks in the soma. Proc Natl Acad Sci U S A 99: 6806-6811.

2. Jelinic P, Stehle JC, Shaw P (2006) The testis-specific factor CTCFL cooperates with the protein methyltransferase PRMT7 in H19 imprinting control region methylation. PLoS Biol 4: e355.

#### FRIDAY 12:30 - 12:45 [F3 MAPPING]

### A Comparative And Integrative Genomics Approach For Identification Of Dairy QTL Candidate Genes

Arun Jayappa<sup>1,\*</sup> Jerry Wei<sup>1</sup> Julie Cavanagh<sup>1</sup> Mehar Katkhar<sup>1</sup> Matthew Hobbs<sup>1</sup> Paul Sheehy<sup>1</sup> Herman Raadsma<sup>1</sup> Peter Williamson<sup>1</sup>

<sup>1</sup>University of Sydney

Keywords: Gene and QTL Mapping Functional Genomics

Integrating genetic and functional genomic data provides a useful approach to identification of candidate genes. The primary objective of this study is to develop an integrative approach for the study of mammary development and lactation. Two different approaches used are, one that focuses on analysis of candidate genes across species and the other on dairy QTL.

Using phenotype and genotype data from approximately 1600 Australian Friesian bulls, association analysis was performed for haplotag SNPs or SNPs and milk protein yield or protein percentage. 15,000 SNPs were first tested in this association. The analysis revealed significant association of a large numbers of SNPs with protein production traits. Reduction of the potential candidate regions was performed using a scoring method that is based on a meta-QTL analysis. Genes derived from association analysis of RNA expression profiling data from mammary tissue was interrogated before integration with QTL candidates. Based on presence call and level of expression or differential expression, a refined list of candidates was generated. The candidates were then analysed for ontology, pathway and gene network interactions. Ongoing refinements to include detailed functional annotation of candidates will be discussed.

#### FRIDAY 12:30 - 12:45 [F2 CONSERVATION GENETICS 1]

An investigation of MHC diversity in koalas (Phascolarctos cinereus) Sarah Elizabeth Jobbins<sup>1,2,3,\*</sup> Claire Elizabeth Sanderson<sup>2,3</sup> Damien Higgins<sup>1,3</sup> Mark Krockenberger<sup>1,3</sup> Katherine Belov<sup>2,3</sup> <sup>1</sup>Koala Infectious Disease Research Group (KIDRG) <sup>2</sup>Australasian Wildlife Genomics Group (AWGG) <sup>3</sup>Faculty of Veterinary Science, University of Sydney Keywords: Population Genetics Conservation Genetics

The koala (Phascolarctos cinereus) is arguably one of Australia's most charismatic icons. Diseases such as Chlamydia and Cryptococcosis are significant causes of koala morbidity and mortality and can amplify population decay in already fragmented habitats. The susceptibility of koalas to disease may be due to an immune system poorly adapted to these pathogens, fairly recent exposure, energy constraints, or a lack of genetic diversity, especially at Major Histocompatibility Complex (MHC) genes. In this presentation we focus on MHC diversity in koala populations. The MHC is a region of the genome containing closely linked genes encoding cell surface proteins that recognize, bind and present pathogen-derived antigens to the immune system. Class II receptors form a gateway to the adaptive immune system: only cells that carry Class II MHC can present antigen to T helper lymphocytes, initiating an adaptive response. The MHC genes are the most polymorphic genes in the vertebrate nuclear genome, making them prime candidates for use in population studies, and can provide information regarding selective processes, the interactions of individuals with their environment and even their capacity for adaptation and evolution. This investigation reports the isolation of the !1 domain of the MHC Class II DAB gene in P. cinereus, characterisation of the amino acid sequence and examination of genetic diversity in 4 populations of koalas. Diversity is compared between outbred, mainland populations in South East Queensland and the mid-North coast of New South Wales and the inbred populations of Victoria's French Island and South Australia's Kangaroo Island. We test the hypothesis that koalas exhibit reduced Class II diversity and find that DAB is a multicopy gene with reasonable diversity in outbred populations. The discovery of a processed MHC pseudogene has raised guestions about retroviral involvement in MHC evolution. The implications will be discussed.

#### WEDNESDAY 14:30 - 14:45 [W5 PHYLOGENY]

#### **Evolution of Optical Microstructures on the Wings of Butterflies**

Shelley Wickham1,2 Julia C Jone<sup>s3,\*</sup> Tiziana Beninati<sup>3</sup> Maryanne CJ Large<sup>2</sup> Leon Poladian<sup>2,4,5,6</sup> Lars S Jermiin<sup>3,5</sup>

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Keywords: Evolutionary Genetics Phylogeny, Biodiversity & Barcoding

Many butterfly species possess 'structural' colour, where colour is due to optical microstructures found in the wing scales. A number of such structures have been identified on butterfly scales. In this study, we characterize the optical properties of different multi-layer structures found across a variety of species. The optical mechanism of suppression and exaggeration of angle-dependent optical properties (i.e., iridescence) of these structures is described. In addition, we consider the phylogeny of the butterflies, and are thus able to relate the optical properties of the structures to their evolutionary development. By applying different phylogenetic principles, we elucidate the evolutionary mechanisms of adaptation. For example, a simple parsimony analysis, in which all evolutionary changes are given equal weighting, implies convergent evolution of some structures; on the other hand, a Dollo parsimony analysis, in which the 'cost' of losing a structure is less than that of gaining it, implies that 'latent' structures can be reused.

WEDNESDAY 14:30 - 14:45 [W6 FUNCTIONAL GENOMICS 1]

The role of a new class of transcriptional activator in the response to nutrient limitation and programmed cell death

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Keywords: Functional Genomics Pathogens, Parasites & Symbionts

We have identified three genes involved in the response to nutrient depletion in the filamentous fungus, Aspergillus nidulans. Two of the genes encode atypical hexokinases (HxkC and HxkD) and the third gene product (XprG) belongs to a newly defined class of DNA-binding proteins. Vib-1, a regulator of genes required for programmed cell death in N. crassa is a homolog of XprG. The XprG/Vib-1 class of proteins show similarity to p53, a regulator of genes involved in cell cycle arrest and apoptosis. The function of a human homolog of XprG has not been determined but it is highly expressed in metastatic tumour cells. xprG- mutants are protease-deficient and have pale conidia suggesting the XprG is involved in regulating extracellular protease and conidial pigment production. To investigate whether XprG is required for expression of other genes in response to carbon starvation, we used microarrays provided by the Pathogen Functional Genomics Resource Center sponsored by NIAID. These experiments indicate that expression of many genes is greatly reduced in an xprG- null mutant. Many of the genes with altered expression (e.g. genes in the sterigmatocystin/aflatxoin biosynthetic pathway) are known to be regulated in response to carbon starvation. These results indicate that XprG may play a major role in the response to carbon limitation.

Genetic evidence indicates that HxkC and HxkD are negative regulators of XprG. We have recently found that unlike HxkD, which is a nuclear protein, HxkC is associated with mitochondria. Mitochondrial hexokinases have been shown to block apoptosis in mammals and programmed cell death in plants. We are investigating whether HxkC plays a similar role in fungi.

#### WEDNESDAY 14:30 - 14:45 [W4 COMPARATIVE GENOMICS 3]

Cytogenetic and Inactivity Map of the Tammar Wallaby X Chromosome

Edda Koina<sup>1,\*</sup> Ruth Doherty<sup>1</sup> Paul D Waters<sup>1</sup> Janine E Deakin<sup>1</sup> Jennifer AM Graves<sup>1</sup> The ARC Centre for Kangaroo Genomics

Keywords: Comparative Genomics Functional Genomics

Comparative genomics is a valuable tool for exploring the organization, function and evolution of the mammalian genome. Comparison of gene location between distantly related species can provide information about genome rearrangement in evolution, and comparison of genetic control can reveal how regulatory mechanisms evolved and function.

Marsupials diverged from eutherians approximately 180 million years ago. Genes on the marsupial X chromosome all map to the long arm of the human X chromosome, defining an X conserved region. The human genes located on this region undergo X chromosome inactivation (XCI). A unique regulatory mechanism that achieves dosage compensation of X linked genes between males and females. It is a whole-chromosome effect under the control of the XIST locus, although some genes escape inactivation. Marsupial XCI differs from the eutherian process: as it is tissue specific, less stable, does not include DNA methylation, is paternal rather than random and does not involve the XIST gene. Raising many fundamental questions about the mechanism and evolution of this important epigenetic system.

As part of the Kangaroo Genome Project we are finalising a cytogenetic map of the Tammar Wallaby (Macropus eugenii) X chromosome. A Tammar Wallaby BAC library has been screened and isolated clones have been mapped to the X chromosome by DNA fluorescence in situ hybridisation (DNA FISH). A number of these isolated gene containing BAC clones are being used in RNA in situ fluorescence hybridisation (RNA FISH) experiments, to create an activity map of the tammar X. Allowing us to distinguish between the hypotheses of local control versus spreading control of XCI over domains on the tammar wallaby X. RNA FISH has been extensively used to investigate gene inactivity in eutherian mammals, as it is extremely sensitive, allowing visualization of RNA at the site of transcription in individual nuclei.

#### WEDNESDAY 14:00 - 14:15 [W4 COMPARATIVE GENOMICS 3]

**The Kangaroo Genome - Filling The Phylogenetic Gap** Elizabeth Kuczek<sup>1,\*</sup> Annette McGrath<sup>1</sup> Peter Wilson<sup>1</sup> Artem Men<sup>1</sup> Daniel Thomas<sup>1</sup> Lankesha Yapa<sup>1</sup> Janette Edson<sup>1</sup> Tanya Levchenko<sup>1</sup> Mehlika Hazar-Rethinam<sup>1</sup> Carmen Troon<sup>1</sup> Allison Hall<sup>1</sup> Amber Stephenson<sup>1</sup> John Davis<sup>1</sup> David Wood<sup>1</sup> Sarah Williams<sup>1</sup> Yogi Sundaravadanam<sup>1</sup> George Weinstock Richard Gibbs<sup>2</sup> Sue Forrest<sup>1</sup>

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Keywords: Comparative Genomics Bioinformatics Evolutionary Genetics

The genome of the model Australian kangaroo Macropus eugenii (the tammar wallaby) has been sequenced to 2x coverage. It is representative of a mammalian species that is 100 million year divergent from placental mammals and 70 million years divergent from the opossum, Monodelphis domestica, the only other marsupial genome sequence that is available.

There are 270 species of marsupials, of which 200 are found in Australia. The tammar wallaby, a small kangaroo species and member of the largest family of Australian marsupials, the Macropodidae displays unique biological features of long standing research interest. These include: arrested embryonic development (diapause), mammary glands elaborating milk of different compositions to support the growth and development of two siblings of different ages. In addition, wallabies are born 27 days after conception before hindlimbs or gonads have developed with embryonic kidney, immune and reproductive systems but functional respiratory, circulatory and digestive systems. They complete their development over the next 9-10 months in their mother's pouch. This offers novel opportunities to study key mammalian developmental processes.

Initial analysis is underway to explore the genome organisation of this marsupial that occupies an exclusive phylogenetic position between placental mammals and birds/reptiles.

Further syntenic genome analyses will provide clues for elucidating the biological distinctiveness of marsupials and mammals as well as a wider comparative genome analysis with the monotreme species, the platypus and echidna, two other unique residents of the Australian continent.

#### FRIDAY 11:15 - 11:30 [F1 POPULATION GENETICS 2]

### Genetic Diversity in Laticaudid sea kraits : Is philopatry resulting in genetic **divergence?** Amanda Lane<sup>1,\*</sup> Richard Shine<sup>1</sup>

University of Sydney

Keywords: Population Genetics Speciation and Phylogeography

Two species of sea kraits, Laticauda laticaudata and Laticauda colubrina, have been found to exhibit significant philopatry, such that they return to the same island repeatedly, even when translocated several kilometres away. Unlike true sea snakes, which are completely aquatic and give birth to live young, Laticaudine seas kraits are amphibious and oviparous, laying their eggs on land. This may be a potentially fragmenting factor in their distribution as breeding sites are not continuously distributed

and a lack of gene flow between island populations may be resulting in genetic divergence. This idea of population fragmentation and accompanying intra-specific divergence is supported by observations of variation in morphological traits, colour and sexual size dimorphism between island populations. My study uses maternally and bi-parentally inherited genetic markers to examine population structure and dispersal in the Laticauda genus. I address whether islands in the central Pacific Ocean maintain genetically distinct populations of sea kraits and whether these species exhibit sex-biased dispersal. Fine scale analyses of both species indicate that populations are not strictly delineated according to their island of capture, although differences do exist at a larger geographic scale. The typical reptilian pattern of male-biased dispersal is not found in the Laticauda genus. Exceptionally high levels of linkage disequilibrium suggest that population structuring is present, however it is not related to the island of capture. An investigation of the relationship between this hidden population structure and the geographic features of the area is underway, as is sequencing of the ND4 region of the mitochondrial genome.

#### WEDNESDAY 11:15 - 11:30 [W3 BIOINFORMATICS] **Compositional Heterogeneity and Model Misspecification in Phylogenetic Studies** Kwok Wai Lau<sup>1,\*</sup> Michael A. Charleston<sup>1</sup> Lars S Jermiin<sup>1</sup> <sup>1</sup>University of Sydney **Keywords:** Bioinformatics Phylogeny, Biodiversity & Barcoding

Markov models are used in many phylogenetic studies of aligned sequence data to approximate the true, but unknown, evolutionary process. Several Markov models exist for both DNA and protein. Choosing the appropriate Markov model entails finding the Markov model(s) that provide(s) the best fit, given the alignment and the tree. The commonly used model-selection methods in phylogenetics are the hierarchical Likelihood Ratio Test (hLRT) and the Akaike Information Criterion (AIC). The two methods have well described advantages and disadvantages; as such, there is a tendency to use the least-constrained model-selection method (i.e., the AIC).

We have surveyed the performance of the hLRT and AIC using DNA generated using Monte Carlo simulations. We discovered that when the sequences evolved under time-reversible conditions (i.e., under the stationary, reversible and homogeneous condition), the method based on hLRT was more likely than that based on AIC to identify the correct Markov model, and that the latter method tends to select more complex time-reversible Markov models than the method based on hLRT. This result is consistent with those published by David Posada (J. Mol. Evol. 52, 434-444). We also discovered that when the sequences evolved under more complex conditions, the two model selection methods performed poorly. In general, both methods identified time-reversible Markov models as the most appropriate approximations of more general evolutionary processes (e.g., leading to compositional heterogeneity). Given the wide spread occurrence of compositional heterogeneity in phylogenetic data, there is a good reason to be concerned about the used of current implementations of model selection methods.

#### THURSDAY 12:15 - 12:30 [TH2 EVOLUTIONARY GENETICS]

## Did one of the lineages of Pediculus (lice) evolve on Homo neanderthalensis? Are the head lice and body lice of humans the same or different species?

Natalie Leo<sup>1,2,\*</sup> Takashi Kageyama<sup>2</sup> Stephen Barker<sup>1</sup>

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Keywords: Speciation and Phylogeography Evolutionary Genetics

Previous phylogenetic studies showed that the head lice and body lice of humans are not reciprocally monophyletic; this has been interpreted as evidence that head lice and body lice are the same species. But here we report new evidence from phylogenetics that head lice and body lice are different species. We studied a mitochondrial gene and a nuclear gene. Genes from individual head lice were often in the Head+Body lice gene-tree (H+B gene-tree) for the mitochondrial gene but in the Head lice only gene-tree (H-only gene-tree) for the nuclear gene, and vice versa. This indicates that different lineages of head lice have interbred and that the gene-trees, of the mitochondrial gene and the nuclear gene we studied, are not congruent with the species-tree of the head lice and body lice of humans. In contrast to head lice, both the mitochondrial and the nuclear genes of individual body lice were always in the H+B gene-tree; this is evidence that body lice have not interbred with head lice from the H-only lineage. Furthermore, our data indicate that the H+B gene-trees initially evolved in lice that were living on

Homo sapiens in East Africa whereas the H-only gene-trees initially evolved in lice that were living on Homo neanderthalensis in Europe. Here we propose that head lice with genes in the H+B genetrees began to interbreed with head lice that had genes in the H-only gene-trees when H. sapiens and H. neanderthalensis came into physical contact in Europe. According to our new hypothesis, head lice and body lice were distinct species when H. sapiens and H. neanderthalensis came into contact, tens of thousands of years ago.

#### FRIDAY 12:30 - 12:45 [F1 POPULATION GENETICS 2]

### The Y chromosome as a tool in populations genetics and disease association studies Joanne Maree Lind<sup>1,\*</sup>

<sup>1</sup>School of Medicine, University of Western Sydney

Keywords: Population Genetics Evolutionary Genetics Gene and QTL Mapping

The non-recombining region of the Y chromosome is unique for association and mapping studies as it is haploid, is present only in males, and genetic variation is not created by recombination. Any single nucleotide change that arises within this region of the Y chromosome will be passed onto all the descendents of the individual in which it arose. This enables males to be assigned to a Y haplogroup by the state (ancestral or derived) of a combination of single nucleotide polymorphisms. The aim of this paper is to demonstrate how Y chromosome haplogrouping can be applied to population genetics and disease association studies. Work will be presented showing how Y chromosome haplogroups, in conjunction with mitochondrial DNA haplogroups, can be used to estimate the relative contribution of parental genomes in admixed populations. Sex biased gene flow can also be determined by this method as demonstrated in African Americans who have a threefold higher European male genetic contribution compared with European females. Y chromosome haplogroups are also useful in disease association studies.

WEDNESDAY 11:45 - 12:00 [W3 BIOINFORMATICS]

CpG substitution rate variation in mammalian genomes

Helen Lindsay<sup>1,\*</sup> Gavin Huttley1

<sup>1</sup>John Curtin School of Medical Research

Keywords: Comparative Genomics Bioinformatics Evolutionary Genetics

Mutations at CpG sites occur much more readily than mutations in other sequence contexts, and contribute significantly to human genetic disease. The nucleotide substitution rate at CpG sites is known to vary with regional nucleotide composition. We have investigated the reasons underlying this variation by examining regional variation in the substitution rate of other sequence contexts. Novel models of dinucleotide substitution have been developed to distinguish substitution patterns caused by the influence of specific sequence contexts from those caused by the properties of individual nucleotides. We found two distinct patterns of regional variation in context dependent substitution rates. The nearest neighbours of a substituted nucleotide predicted which type of pattern it exhibited. Specifically, nucleotides with a C or G neighbouring nucleotide exhibited the highest substitution rate in G+C rich regions and the lowest substitution rate in A+T rich regions. The converse was true for nucleotides with an A or T neighbouring nucleotide. After accounting for these neighbouring nucleotide effects, we found that regional variation in the CpG substitution rate was greatly reduced. We propose that the neighbouring nucleotide effect originates from a nearest neighbour influence on the DNA polymerase extensionproofreading equilibrium. If so, regional variation in the CpG substitution rate may derive from regional variation in the repair of lesions at CpG sites rather than from regional differences in the rate of lesion formation.

THURSDAY 12:45 - 13:00 [TH2 EVOLUTIONARY GENETICS]

**Reduced microsatellite variation on a marsupial Y chromosome** Anna J MacDonald<sup>1,\*</sup> Stephen D Sarre<sup>1</sup> Nancy N FitzSimmons<sup>1</sup> <sup>1</sup>University of Canberra

Keywords: Evolutionary Genetics Population Genetics

A thorough understanding of the mutational mechanisms acting on microsatellite DNA, especially those caused by recombination, is essential to accurately interpret microsatellite genotyping data. In marsupials the Y chromosome does not undergo any recombination, while the X chromosome can recombine in XX females but not in XY males. Tammar wallaby (Macropus eugenii) samples were genotyped at 29 microsatellites originating from the X and Y chromosomes and chromosome 2. Allelic richness was positively correlated with repeat length. Controlling for repeat length, allelic richness was significantly lower on the Y chromosome than on chromosome 2, with the X chromosome intermediate. Reduced microsatellite diversity on the Y chromosome may reflect fewer

mutations on the Y chromosome due to the lack of recombination. Alternatively, reduced Y chromosome diversity could be a consequence of demographic factors or the lower effective population size of the Y chromosome relative to autosomes. Smallpool PCR from sperm is currently being explored as a method to identify the types of mutations occurring at tammar wallaby microsatellites. This may provide further evidence for the role of recombination in the microsatellite mutation process. The discovery of reduced Y chromosome diversity in a wallaby may also need to be considered in the context of conservation management for this and related species.

#### FRIDAY 11:00 - 11:15 [F2 CONSERVATION GENETICS 1]

#### Effects of an invasive toxic prey on the population genetics of a naïve native squamate predator: cane toads and goannas in the Northern Territory

Thomas Madsen<sup>1,\*</sup> Beata Ujvari<sup>1</sup>

<sup>1</sup>School of Biological Sciences, University of Wollongong Keywords: Conservation Genetics Population Genetics

Human introduction of numerous exotic animal and plant species into the Australian continent has often had catastrophic effects on the indigenous fauna and flora. A fairly recent introduction was the release of the South American cane toad (Bufo marinus) into the sugar cane fields of Queensland in 1935. The cane toad is one of the most toxic bufonids and when seized and swallowed by naïve Australian predators the toxin will usually result in the demise of the attacker. One group of Australian squamate reptiles that has been suggested to be very susceptible to cane toad toxin is varanid lizards. Prior to the invasion of cane toads into our study area, situated in the Top End of the Northern Territory of Australia, annual mortality of adult male radio-tracked Varanus panoptes was very low (two deaths recorded among 20 lizards over three years). By contrast after the arrival of the toads in October 2005, all of radio-tracked goannas were found dead in August 2006 (nine out of nine lizards). Our results suggest that the vast majority, most likely > 90%, of the naïve adult male goannas will succumb when encountering cane toads. Such a significant increase in mortality will result in a substantial reduction in goanna population genetic diversity. Our work on small, bottle-necked squamate reptile populations in Europe has demonstrated that loss of genetic diversity being the most significant factor resulting in a substantial increased risk of extinction. We therefore suggest that in order to ensure the future long-term survival of the remaining small goanna populations warrants intensive population genetic studies, possibly involving genetic rescue.

#### FRIDAY 11:15 - 11:30 [F3 MAPPING]

**QTL mapping for heavy metal tolerance in chironomids** Cintia M. Martinez-Garduno<sup>1,\*</sup> Yvonne M. Parsons<sup>1</sup>

<sup>1</sup>Department of Genetics and Human Variation, La Trobe University, Victoria, 3086, Australia Keywords: Gene and QTL Mapping Adaptation

Chironomids (non-biting midges) are abundant, widespread and useful invertebrates for biomonitoring of aquatic ecosystems. Biomonitoring is commonly used for environmental assessment and has proved to be more informative than traditional physical-chemical analyses. The incorporation of biomarkers can provide information on toxicant exposure in an organism at the genetic level and has the potential to be an early-warning indicator for water health. Biomarkers can be developed when organisms can tolerate specific environmental stresses such as heavy metals or pesticides, as is the case with chironomids. Tolerance to heavy metals in chironomids has been suggested to be due to the induction of metal binding proteins such as methallothioneins, or heat shock proteins. However, the genetic basis of this response is still poorly understood.

We are investigating the basis of heavy metal tolerance in Chironomus tepperi through QTL mapping. C. tepperi is endemic to south east Australia where it has become a pest of rice crops, tolerating high pesticide concentrations. This suggests it may be a useful model organism to investigate for adaptation to other environmental stresses such as heavy metal exposure. We have previously demonstrated that C. tepperi can develop metal tolerance over four generations of selection and are now in the process of generating lines for QTL mapping. Chironomids are highly suitable for QTL analysis as they can be reared successfully under laboratory conditions, produce numerous offspring from pair wise matings and have a short life cycle.

This study represents the first attempt to identify QTL for heavy metal tolerance in chironomids and will serve as the basis to clarify the genetics underlying this response. This study could also provide useful information for future development of biomarkers to improve biomonitoring in aquatic waterways affected by heavy metal pollution.

FRIDAY 11:30 - 11:45 [F2 CONSERVATION GENETICS 1]

Reduced MHC class II diversity in island compared to mainland populations of a threatened rock-wallaby (Petrogale lateralis lateralis)

Robert A Mason<sup>1,2,\*</sup> Teena L Browning<sup>2</sup> Mark D B Eldridge<sup>1,\*</sup> <sup>1</sup>DNA Lab, Australian Museum <sup>2</sup>Department of Biological Sciences, Macquarie University

Keywords: Conservation Genetics Population Genetics

Many mammal species that are endangered on mainland Australia exist in stable island populations. These island populations have the potential to act as an "ark" in the case of mainland population extinctions. Genetic diversity at fitness-associated loci is integral to the viability of populations. It is thus important to investigate fitness related genetic diversity of island populations before they are considered suitable as reservoirs of their species. This was performed with regard to the black-footed rock-wallaby (Petrogale lateralis), which exists in small fragmented mainland populations in Western Australia and on Barrow Island. The Major Histocompatibility Complex class II locus DAB is associated with fitness, through its role in antigen presentation. Diversity at DAB was assessed using single strand conformation polymorphism (SSCP) and sequencing. The mainland populations displayed moderate levels of allelic diversity (4-7 alleles) despite being small in size and isolated from one another, and contain at least two DAB loci. However, the Barrow Island population displayed low allelic diversity (2 alleles) and high monomorphism in comparison to mainland populations, and probably possess only one DAB locus. Previous studies using microsatellites have also suggested that genetic variation in island marsupial populations is low. This study is one of the first to provide explicit evidence that fitness related genetic diversity in these populations is also low. If this situation is similar across other island populations, it would be far better to concentrate resources upon preserving mainland populations than to rely upon island populations as a conservation resource.

FRIDAY 15:00 - 15:15 [F5 ADAPTATION]

Wolbachia infection and insect activity

Peng Yu<sup>1</sup> John Nielsen<sup>1</sup> Kathryn Real<sup>1</sup> Elizabeth McGraw<sup>1,\*</sup> <sup>1</sup>University of Queensland Keywords: Pathogens, Parasites & Symbionts

Wolbachia pipientis is a naturally occurring endosymbiotic bacterium present in a diverse range of insect hosts. The microbe is thought to manipulate host reproductive biology to its advantage from its primary site of occupation and transmission in the gonads. Wolbachia is also found throughout other host tissues where the effects of the infection are less well understood. In the case of a virulent strain of Wolbachia called popcorn, aging flies appear from qualitative observation to be more sedentary and exhibit jittery forms of movement. This mutant strain is known to aberrantly over replicate and may be causing local damage in nervous or muscle tissue. In addition to characterising some of the effects of this extreme strain on hosts, we also wished to determine if naturally occurring Wolbachia infections in Drosophila melanogaster were affecting locomotion as measured by laboratory assays and field dispersal. Using mark-release-recapture, and laboratory olfactory driven capture assays and tests of locomotory performance it appears that the infected lines are also more sedentary. This work has implications for understanding the way that Wolbachia spreads through host populations and for Wolbachia based insect control strategies currently in development.

FRIDAY 11:30 - 11:45 [F3 MAPPING]

### Towards a Comprehensive Genetic-Linkage Map for the Saltwater Crocodile (Crocodylus porosus)

Lee Miles<sup>1,\*</sup> Travis Glenn2 Sally Isberg<sup>1,3</sup> Pauline Dalzell<sup>4</sup> Peter Thomson<sup>1</sup> Cris Hagen<sup>2</sup> Chris Moran<sup>1</sup> <sup>1</sup>Centre for Advanced Technologies in Animal Genetics and Reproduction (Reprogen), Faculty of Veterinary Science, University of Sydney, Australia

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Keywords: Gene and QTL Mapping

We report the development of more than 250 informative microsatellite markers, and the first stage of construction of a comprehensive microsatellitebased genetic linkage map for the saltwater crocodile (Crocodylus porosus). This map represents the first genetic linkage map in the Order Crocodylia and indeed the first in the Class Reptilia. These markers will be used to map Quantitative Trait Loci (QTL) for economically important selection objectives in farmed saltwater crocodiles. Any QTL identified may then be useful in implementing a Marker-Assisted Selection (MAS) genetic selection

program. High-density genetic maps for QTL identification require many polymorphic markers spaced evenly across the genome. Our map, which we anticipate completing by mid-2007, will contain approximately 300 informative genetic markers selected from more than 600 microsatellites that were originally developed and evaluated. The 600 loci will be evaluated for utility in all extant crocodilian species for future research.

#### FRIDAY 12:00 - 12:15 [F2 CONSERVATION GENETICS 1]

### Bigger is not better: male reproductive success in a captive breeding program for the greater bilby (Macrotis lagotis) Emily J Miller<sup>1,\*</sup> Mark D. B. Eldridge<sup>2</sup> Tony Friend<sup>3</sup> Neil Thomas<sup>3</sup> Nicky Marlow<sup>3</sup> Catherine A

Herbert

<sup>1</sup>University of New South Wales,

<sup>2</sup>Australian Museum,

<sup>3</sup>Department of Environment and Conservation

Keywords: Conservation Genetics Population Genetics

The Greater Bilby (Macrotis lagotis) is the only remaining species of mainland desert bandicoot, and is an iconic species for conservation in Australia. They are burrowing, nocturnal marsupials whose reproductive biology is poorly understood due to their secretive nature. 'Return to Dryandra' (RTD) is a captive breeding program developed by Western Australia's Department of Environment and Conservation (DEC) in 1998, and forms part of the Western Shield program. RTD is located in the Dryandra Woodland, where bilbies have become locally extinct. The breeding program aims to provide stock for reintroduction to other Western Shield fauna reconstruction sites.

The primary objective of this study was to determine the levels of genetic diversity maintained since the captive breeding program was established. The secondary objective was to determine the genetic mating system of the greater bilby and whether body size influenced male reproductive success. In this study, 233 individuals were genotyped using nine polymorphic microsatellite markers. This study shows genetic diversity has remained relatively stable since the colony was established in 1998. In any given year a large proportion of males did not contribute to the gene pool as they did not sire any offspring. Sires and non-sires could not be distinguished from one another based on their body size.

#### THURSDAY 11:00 - 11:15 [TH1 POPULATION GENETICS 1]

A Genetic Marker for the Queensland Fruit Fly in SIT programs

Jennifer Morrow<sup>1,\*</sup> Stuart Gilchrist<sup>1</sup> Sasha Curthoys<sup>1</sup> Marianne Frommer<sup>1</sup> Kathryn Raphael<sup>1</sup> Deborah Shearman<sup>1</sup>

<sup>1</sup>Fruit Fly Research Centre, School of Biological Sciences, University of Sydney. Keywords: Phylogeny, Biodiversity & Barcoding

The Queensland fruit fly, Bactrocera tryoni, is the major pest of commercial fruit in eastern Australia. Its geographic distribution extends from the Northern Territory and Queensland to Victoria, and outbreaks have occurred in South Australia and Western Australia. B. jarvisi, a fruit fly found mostly in northern Australia, is only a minor pest of commercial crops. Morphologically, B jarvisi and B. tryoni are easily distinguished. Sequence analysis of mitochondrial and nuclear genes provides no support for the different subgeneric status of the two species, but the data confirm the morphological indications that these flies are very distinct species.

It is interesting to find, therefore, that the two species will mate in the laboratory to produce fertile offspring and that the F1 hybrids can mate successfully with B. tryoni and B. jarvisi. These observations provide important possibilities for genetically marking a strain of B. tryoni, in order to improve Sterile Insect Technique (SIT) programs.

Currently, SIT is the only non-pesticide option for eradicating B. tryoni on an area wide basis. SIT has proven itself a useful tool against many insect species, including the Mediterranean fruit fly, but requires improvement to reach its full potential in Qfly. One ongoing difficulty is the accurate identification, for monitoring purposes, of sterile and wild flies caught in traps. The current method of marking sterile flies with a fluorescent dye has many problems. A genetic marker is needed to distinguish, by means of a simple molecular test, the sterile release strain from wild flies.

We are marking B. tryoni with the mitotype of B. jarvisi mitochondrial DNA, by crossing B. tryoni males with B. jarvisi females. We will determine if the mitochondria are stably inherited and will attempt to generate a robust factory strain for sterilisation and release.

#### THURSDAY 11:00 - 11:15 [TH3 FUNCTIONAL GENOMICS 2]

Molecular evolution and the transition to viviparity

Bridget Frances Murphy<sup>1,\*</sup> Scott Parker<sup>1</sup> Joanna Biazik<sup>2</sup> Katherine Belov<sup>3</sup> Michael B Thompson<sup>1</sup> School of Biological Sciences, University of Sydney

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Keywords: Comparative Genomics Evolutionary Genetics

The evolutionary shift from oviparity (egg-laying) to viviparity (live-bearing) constitutes the most dramatic transition in the reproduction of amniotes. Squamate reptiles are used to investigate this transition because they exhibit many independent evolutionary origins of viviparity. The majority of studies focus on morphological features and physiological or ecological correlates associated with the evolution of viviparity. Instead, we are taking a genetic approach to identify molecular mechanisms involved in some of the major physiological changes involved in the transition to viviparity. We are quantifying uterine expression of four genes at different stages of pregnancy in oviparous and viviparous lizards, with the aim of determining if they are responsible for changes involved in the transition to viviparity. Genes of focus are: 1) hypoxia-inducible factor 1 alpha (HIF-1") and vascular endothelial growth factor because they are probably responsible for proliferation of blood vessels in the uterus of viviparous lizards to deliver oxygen to the embryo during its prolonged gestation; 2) claudins, a family of tight junctional proteins that regulate paracellular transport through epithelia, because they may be associated with the transition from lecithotrophy (nutrients from yolk) to placentotrophy (nutrients across the placenta); and 3) major histocompatibility complex (MHC) Class I genes, responsible for immune recognition of foreign antigens in vertebrates, which may be expressed differently in egg-laying and live-bearing species because viviparous lizards lack the thick eggshell that separates the uterus from the allogeneic embryo in oviparous species. So far, candidate HIF-1", VEGF and MHC Class I genes have been identified in four species of lizards, Saiphos equalis, Eulamprus tympanum, Ctenotus taeniolatus and Pseudomoia entrecasteauxii.

THURSDAY 12:30 - 12:45 [TH1 POPULATION GENETICS 1]

## Population Genetics and Breeding System of the Giant Gippsland Earthworm Megascolides australis

Rebecca Wood<sup>1</sup> David Runciman1 Neil Murray<sup>1,\*</sup> <sup>1</sup>La Trobe

#### Keywords: Conservation Genetics Population Genetics

The Giant Gippsland Earthworm *Megascolides australis* (GGE) is one of the largest known terrestrial invertebrates. Its totally subterranean habit means that little is known of its general biology and breeding behaviour. Because of vegetation clearance and agricultural land management the species is now of conservation concern, being listed as Vulnerable by both IUCN and EPBC. A major road realignment project has led to the opportunity for an experimental translocation of a GGE population. As part of this project, genetic analyses have investigated the genetic structure of GGE populations. Important questions are how genetically distinct different sub-populations are, and whether reproduction is exclusively sexual. Samples from two subpopulations, Loch Hill and Bena, have been analysed. Mitochondrial DNA (COI) sequencing shows that there is considerable variability within subpopulations have been large and isolated for a considerable time. Microsatellite genotypes also indicate high variability both within and between sub-populations. Multilocus genotypes give no evidence of any clonal component of reproduction. Intriguingly, the simplest interpretation of microsatellite profiles is that they represent a relatively recent episode of tetraploidy in GGE ancestry. Implications of these results for conservation management will be discussed.

WEDNESDAY 15:00 - 15:15 [W5 PHYLOGENY]

### Screening For Porcine Endogenous Retrovirus Sequences Among Species Of The Family Suidae

Fabrícia Nascimento<sup>1,\*</sup> Jaime Gongora<sup>1</sup> Stewart Lowden<sup>2</sup> Hans Klingel<sup>3,4</sup> Olivier Hanotte<sup>5</sup> Daniel White<sup>6</sup> Bradley Cain<sup>7</sup> Andreas Spoetter<sup>8</sup> Chris Moran<sup>1</sup>

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<sup>8</sup>Hannover School of Veterinary Medicine, P.O.Box 71 11 80, D-30545 Hannover, Germany **Keywords:** Pathogens, Parasites & Symbionts

Endogenous retroviruses (ERV) are integral components of the host genome resulting from infection of germ line cells. Such proviruses are transmitted vertically. They accumulate mutations and often no longer encode an infectious virus. In pigs, some tissues have been shown to express retroviral RNA and the discovery of PERVs capable of infecting human cells in vitro challenged the safety of xenotransplantation, increasing research interest in PERVs because of their potential zoonotic risk. The study of the relationships of Suid ERVs and their hosts will help in understanding their diversity and coevolutionary patterns of hosts and parasites. This study follows work from this laboratory focused on the the pol and env genes. Here, we use new primers to amplify and sequence bigger fragments of the pol (~1578bp) and env (~2283bp) genes and for the first time ever the gag (~1,084bp) gene in nine species and one subspecies of the Suidae family, namely the babirusa (Babyrousa babyrussa), the forest hog (Hylochoerus meinertzhageni), the common warthog (Phacochoerus africanus), the red river hog (Potamochoerus porcus), the bush-pig (Potamochoerus larvatus), the bearded pig (Sus barbatus oi and Sus barbatus barbatus), the Celebes warty pig (Sus celebensis), the wild boar (Sus scrofa) and the Javan warty pig (Sus vertucosus). Retroviral fragments have been amplified from almost all species of Suidae, but not from babirusa, suggesting either that these ERVs infected the Suidae family after divergence from babirusa, the most basal species, or alternatively that the more ancient divergence of the babirusa has allowed accumulation of mutations in regions complementary to the PERV primers preventing amplification of PERV amplicons from this species. Further analysis will be carried out in order to develop a better understanding of the distribution of PERVs fragments in the Suidae family and establish coevolutionary patterns that might be present in this family.

#### FRIDAY 11:45 - 12:00 [F1 POPULATION GENETICS 2]

**Occurrence of Hybridisation between Sympatric Populations of Grey Kangaroo** Linda E Neaves<sup>1,\*</sup> Kyall R Zenger<sup>2</sup> Mark D. B Eldridge<sup>3</sup> Desmond W Cooper<sup>4</sup> <sup>1</sup>Macquarie University, <sup>2</sup>The University of Sydney, <sup>3</sup>Australian Museum, <sup>4</sup>University of New South

Wales

#### Keywords: Speciation and Phylogeography Population Genetics

The extent and impact of introgressive hybridisation on the evolution of animal taxa has only recently become apparent with improvements in the sensitivity of detection techniques. However, among mammalian species hybridisation appears relatively rare. Whilst hybridisation under captive conditions has been observed in several macropod marsupials there has been little direct evidence for its occurrence in sympatric populations. In this study we investigate the occurrence of hybridisation between sympatric populations of western grey (Macropus fuliginosus) and eastern grey (Macropus giganteus) kangaroo. Twelve autosomal microsatellite loci, displaying frequency shifts between the two species, a section of the mitochondrial DNA (mtDNA) control region and four Y-chromosome microsatellite loci were examined in 223 grey kangaroos. Fifteen percent of the sampled individuals displayed some level of introgression at autosomal microsatellite loci, though no first generation hybrids were apparent. Although hybridisation in captivity appeared unidirectional introgression of autosomal microsatellite alleles occurred in both directions. Additionally, four M. giganteus individuals displayed introgression of M. fuliginosus Y-haplotypes and one case of M. fuliginosus mtDNA introgression was also apparent. Reproductive isolation in grey kangaroos is incomplete and rare instances of hybridisation and subsequent backcrossing have occurred since secondary contact took place, approximately 60,000 years ago. The use of molecular techniques has revealed that despite the unidirectional nature of captive hybridisation and infertility of F1 male hybrids, hybridisation, occurs in wild sympatric populations of grey kangaroo. Hence, the rarity of hybridisation among mammalian species may be more reflective of past difficulties, than biological reality.

#### WEDNESDAY 14:15 - 14:30 [W5 PHYLOGENY]

# DNA barcoding forensically and medically important Australian Chrysomya (Diptera: Calliphoridae)

Leigh A Nelson<sup>1,\*</sup> James F Wallman<sup>2</sup> Mark Dowton<sup>1</sup>

<sup>1</sup>Institute for Biomolecular Science, School of Biological Sciences, University of Wollongong <sup>2</sup>Institute for Conservation Biology, School of Biological Sciences, University of Wollongong **Keywords:** Phylogeny, Biodiversity & Barcoding Population Genetics

A DNA barcoding approach shows potential to be advantageous for the identification of taxa for which the use of morphology, or the association of different life stages, is problematic. For these reasons, we tested the efficacy of cytochrome oxidase I (COI) DNA barcodes for the identification of forensically important blowflies of the genus Chrysomya (Diptera: Calliphoridae), collected from the east coast of Australia. The identification of several Chrysomya species is hampered by their similar morphologies, even as adults. A 658 bp fragment of the COI gene was sequenced from 56 specimens, representing all nine Australian Chrysomya and three calliphorid outgroups. The second ribosomal transcribed spacer (ITS2) was sequenced from some species to verify results obtained using COI. The COI Sequences divergences were calculated using the Kimura-two-parameter distance model, and a bootstrap neighbour-joining (NJ) tree was generated to provide a graphic display of the patterns of divergence among the species. We found the COI barcode to be successful for the identification of Australian Chrysomya, with all species resolved as reciprocally monophyletic groups on the NJ tree, with strong bootstrap support. The only exception was a specimen identified as Ch. saffranea, which was recovered with its sister species, Ch. megacephala, on the NJ tree. Further morphological and molecular evidence led to the conclusion that the specimen was a hybrid, and was not included in further analyses. Intraspecific sequence divergences averaged 0.097% (range = 0-0.612%), while interspecific divergences averaged 6.499% (range = 0.458-9.254%). The overlapping sequence divergences for the Chrysomya is attributable to the low sequence divergence (mean = 0.484%) between Ch. megacephala and Ch. saffranea. We found no difference in species delineation when we compared the NJ tree with more complex Bayesian analyses of the dataset.

#### TUESDAY 11:30 - 11:45 [T1 COMPARATIVE GENOMICS 1]

#### Evolution of sex chromosomes in snakes

Denis O'Meally<sup>1,\*</sup> Tariq Ezaz<sup>1</sup> Steve Sarre<sup>2</sup> Arthur Georges<sup>2</sup> Jenny Graves<sup>1</sup>

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<sup>2</sup>Institute for Applied Ecology, University of Canberra, Bruce, ACT Keywords: Comparative Genomics Evolutionary Genetics

Sex determination is accomplished in a great variety of ways, including chromosomal (XY and ZW systems), genetic (GSD) and environmental sex determination (ESD). How these systems relate to each other has been a matter of scientific debate for more than a century. A conserved sex determining gene and conserved sex chromosomes seem unlikely in vertebrates, yet the sex chromosomes of birds and snakes (which are not closely related) appear superficially almost identical. Sex determination is a complex process and the most important developmental decision for an individual. The study of sex chromosomes and the genes they hold is the basis for understanding this fundamental process. We are using a variety of reptile species to study the genetic relationships of their sex chromosomes and the genes involved in sex determination. Broadly our aims are to i) discover the molecular and cytological relationships of sex chromosomes among reptiles ii) discover the ancestral chromosome pair(s) that became sex chromosomes in birds and snakes iii) discover novel sex determining genes in reptile and bird sex determining pathways. We are using repetitive DNA elements to investigate the relationships of the heteromorphic sex chromosome in birds and snakes. We have discovered a novel class of repeats common to the chicken and snake W, though their accumulation on the W in the derived lineages of snakes is greatly advanced. We are investigating the cytological distribution of the banded krait minor (Bkm) satellite repeat among snakes. Its differential accumulation on the Z and W chromosomes makes it a candidate for a PCR based molecular sexing test. We have developed the first such molecular test for sex in snakes.

#### FRIDAY 11:00 - 11:15 [F3 MAPPING]

Mapping the Gene for Worker Sterility in the Honey Bee (Apis mellifera) Peter Robert Oxley<sup>1,\*</sup> Ben P Oldroyd<sup>1</sup> Nathan Lo<sup>1</sup> <sup>1</sup>University of Sydney Keywords: Gene and QTL Mapping Adaptation

Despite the importance that genes have to our theoretical understanding of social evolution, few empirical studies have attempted to isolate genes that underpin natural variation in social traits. In honey bees, a truly social insect, workers are characteristically sterile. Finding the genes that suppress reproduction in this all-female caste is an immediate goal of sociobiology.

Using genomic resources available for the honey bee, and a purpose bred strain of worker (anarchist) expressing high levels of ovary activation, we have established a backcross population that segregates for an unknown gene strongly affecting ovary activation. Using this population we have applied microsatellite-based quantitative trait loci (QTL) mapping to 97% of the genome.

This screen has revealed a single significant QTL accounting for 15% of the variation in ovary activation rates observed in our backcross population. This QTL is believed to be a secondary locus involved in the ovary activation pathway. Examination of this QTL has revealed a list of positional candidates, including two odorant receptors and a dopamine receptor.

#### THURSDAY 12:00 - 12:15 [TH3 FUNCTIONAL GENOMICS 2]

Expression of Myostatin in Cattle Selected for High and Low Muscling and in High Muscling Cattle Heterozygous for a Myostatin loss of Function Mutation

Grant Peter Parnell<sup>1,2,\*</sup> Paul Leo Greenwood<sup>1,3</sup> Yizhou Chen<sup>1,</sup>

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Keywords: Functional Genomics

This paper will investigate differences in myostatin gene expression in muscle tissue of cattle selected for high and low muscling and in high muscling cattle that are heterozygous for the myostatin loss of function mutation (nt821del11). NSW DPI has established high and low muscling selection lines of Angus cattle which vary in retail beef yield. Within the high muscling line cattle, there are individuals that are heterozygous for the loss of function mutation for myostatin. This has been shown to further increase retail beef yield by a further 3.5% (Café et al, 2006).

The most powerful genes for increased muscling in cattle are associated with mutant forms of the myostatin gene (GDF8) which occur in breeds such as Piedmontese and Belgian Blue (Arnold et al, 2001). In its functional form, the myostatin gene produces a protein that is a negative regulator of muscle growth and development. It is known to influence stem cell differentiation and inhibit prenatal myoblast differentiation and growth (Arnold et al, 2001). In its mutant form, the myostatin gene acts as a switch that favours myogenesis at the expense of adipogenesis. However, the extent to which differences in gene expression between cattle that differ in muscling are specifically associated with the myostatin gene, or whether other genes associated with selection for muscling may contribute to these findings, is unclear.

Myostatin gene expression will be determined using a quantitative real-time PCR thermocycler that allows high resolution melt curve analysis to be carried out. This enables differentiation between alleles of the myostatin gene. Results will show the different levels of myostatin expression in Longissimus dorsi and Semitendinosus muscles in the three groups of animals.

Arnold H, Della-Ferra M, Baile C 2001, Review of myostatin history, physiology and applications, International Archives of Bioscience, 2001 pp 1014-1022

Café L, O'Rourke B, McKiernan W, Greenwood P 2006, Carcass and yield characteristics of steers from Angus muscling selection lines with normal and mutant myostatin alleles, Australian Society of Animal production 26th Biennial Conference Short Communication

#### TUESDAY 18:55 - 19:10 [S1 STUDENT]

### Characterisation of the beta-globin cluster and its flanking regions in the egg-laying monotreme, Ornithorhynchus anatinus (platypus)

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Keywords: Comparative Genomics Bioinformatics Evolutionary Genetics

The alpha and beta globin gene clusters contain genes that together encode the polypeptide chains of haemoglobin. Although these genes have been extensively studied in vertebrates, questions about their evolution still remain. In humans, there are five active !-like globin genes in the  $\beta$ - cluster linked in the order 5'- $\epsilon$ - $\gamma$ G- $\gamma$ A- $\delta$ - $\beta$ -3'. They are arranged in their order of expression,  $\epsilon$  (embryo),  $\gamma$  (fetus) and  $\delta$ - $\beta$  (adult). Marsupials contain three beta-like genes;  $\epsilon$  and  $\beta$  (5'- $\epsilon$ - $\beta$ -3') located in the  $\beta$ -cluster, as well as the marsupial-specific & located adjacent to the  $\omega$ -cluster. We have isolated and characterised the  $\beta$ -like globin genes in a monotreme species, the platypus (Ornithorhynchus anatinus), in order to investigate the molecular evolution of the gene family in mammals. Platypus genomic BAC libraries were screened and the isolated BACs were localized onto platypus chromosomes. The sequences of these BAC clones were analysed to fully characterise genes contained in them. These results show that platypus contains three  $\beta$ -like globin genes, one encoding an amino acid sequence identical to the previously isolated adult  $\beta$ -globin protein of the platypus. Another is similar to other mammalian embryonic  $\epsilon$ -globin gene proteins, but phylogenetically groups with monotreme adult  $\beta$ -globin genes, and the third is similar to the marsupial  $\omega$ -globin gene. Cytogenetic mapping showed that the  $\beta$ -cluster, excluding the  $\omega$  gene, is located on the long arm of the platypus chromosome 2 (2q5.1).

Phylogenetic footprinting identified olfactory genes that embed the  $\beta$ -globin cluster (also in therian mammals) and a regulatory region called the locus control region (LCR) that is likely to regulate the differential expression of  $\beta$ -like globin genes in platypus. The presence of a globin gene similar in structure to embryonic  $\varepsilon$ -globins and located 5' to the adult  $\beta$ -globin but phylogenetically grouping with adult  $\beta$ -globins can be explained by two different hypotheses. In the first hypothesis, the ancestral  $\beta$ -globin gene duplicated just prior to the monotreme-therian divergence, with gene conversion events or stochastic lineage sorting resulting in the monotreme  $\varepsilon$ -globin phylogenetically grouping with monotreme  $\beta$ -globin. Alternatively, the  $\varepsilon$ -globin gene has evolved twice, once from a duplication within the monotreme lineage and again by duplication after the monotreme-therian divergence.

WEDNESDAY 14:45 - 15:00 [W6 FUNCTIONAL GENOMICS 1]

#### Molecular factors involved in Thielaviopsis basicola-plant interactions

Lily Pereg-Gerk<sup>1,\*</sup> Samiya Al-Jaaidi<sup>1</sup> Margaret Katz<sup>1</sup>

<sup>1</sup>University of New England

Keywords: Pathogens, Parasites & Symbionts Pathogens, Parasites & Symbionts

Black root rot presents a substantial threat to crops, such as cotton, tobacco, lettuce, pansy, red clover, carrots, strawberries and cucumbers. The disease, caused by the fungal pathogen *Thielaviopsis basicola*, is a significant threat to crops in Australia, especially in cooler areas and seasons. The soilborne fungal pathogen produces thick walled spores that can survive in the soil for years. While management strategies based on cultural practices can reduce the severity of the disease, there is considerable scope for new disease control methods based on an improved knowledge of the biology of the pathogen and its interactions with cotton. One of the strategies we use to study the infection process is the generation of mutants altered in the ability to complete one or more of the main six steps in establishing infection, identify the genes/proteins involved and their control mechanisms. Our aim is to explain why some fungal-root interactions are compatible, leading to disease, and others are incompatible, leading to the phenomenon of resistance.

We develop methods for random mutagenesis (integrative transformation) of *T. basicola*, including PEG-mediated transformation, using fungal protoplasts, and *Agrobacterium tumefaciens*-mediated transformation. We have managed to transform *T. basicola* with plasmid pGpdGFP using PEGmediated transformation, and so far isolated five mutants affected in pathogenicity (out of 208 transformants). Southern blot analysis revealed random insertion of the plasmid into the fungal genome, indicating possibly five different mutations. The mutants differ in several phenotypical characteristics, but the genes affected still need to be rescued. Alternative methods are being tested for transformation, which would simplify the rescue and identification of the mutated genes in the future. The plant response to *T. basicola* pathogenicity mutants will be tested in a related project involving cotton proteomics.

#### WEDNESDAY 15:30 - 15:45 [W6 FUNCTIONAL GENOMICS 1]

### Generation and characterisation of resistance to neonicotinoids in Drosophila

#### melanogaster

Trent Perry<sup>1,\*</sup> John McKenziel Philip Batterham<sup>1</sup>

<sup>1</sup>CESAR - Department of Genetics, Bio21 Institute, The University of Melbourne **Keywords:** Functional Genomics Adaptation

Resistance to insecticides by modification of their molecular targets is a serious problem in the chemical control of many arthropod pests. Neonicotinoids target the nicotinic acetylcholine receptor (nAChR) of arthropods for which the spectrum of possible resistance-conferring mutations is poorly understood. Prediction of these is complicated by the existence of ten nAChR subunit genes, some of which are capable of a generating a number of splice isoforms.

Guided by an initial resistance screen on deficiencies of different nAChR subunits, we focused on the cluster of three *Drosophila melanogaster* nAChR subunit genes at cytological region 96A. EMS mutagenesis and selection for resistance to nitenpyram in hybrids carrying a deficiency for this chromosomal region identified four recessively resistant mutants. Complementation analysis identified two groups, and mutations were found in two different nAChR subunit genes,  $D\alpha I$  (encoding an  $\alpha$ -type subunit) and  $D\beta 2$  ( $\beta$ -type).

Mutations conferring resistance in  $\beta$ -type receptors have not previously been reported and we found a variety of different lesions in the predicted D $\beta$ 2 protein, including locations distant from the residues predicted to be involved in the insecticide binding site. This work clearly demonstrates that mutations in a single receptor subunit can confer neonicotinoid resistance which was not clear in previous studies.

Interestingly, mutations were found that may protect the insect against nitenpyram by interfering with subunit assembly or channel activation, rather than blocking binding of insecticide to the channel and also two nonsense mutations were found which would result in non-functional subunits. This has parallels to studies on spinosad resistance, where the knockout of nAChR subunit  $D\alpha 6$ , confers high levels of resistance. The ability of an insect to survive without particular subunits and the redundancy of the insect nervous system are issues worth considering in future insecticide design.

#### THURSDAY 11:45 - 12:00 [TH3 FUNCTIONAL GENOMICS 2]

A Comparative Genomics Aproach To Identify Lactation Candidate Genes Palaniappan Ramanathan<sup>1,\*</sup> Rosanne Taylor1 Peter Williamson1 <sup>1</sup>University of Sydney

Keywords: Comparative Genomics Functional Genomics

The extensive use of mouse models for cell biology and mammary gland research has generated a wealth of information that may be directly related to lactation. Comparative genomics is a powerful tool for exploiting the mouse genome database to find genes of interest underlying traits of interest in the dairy cattle. Inbred strains of mice differ greatly in their lactation performance and may serve as good models for identification of putative candidate genes. A strain of mice (QSi5), developed at the University of Sydney has demonstrated a superior lactation performance when compared to other mouse strains. Comparison of gene expression profiles in this strain with the CBA strain will help in identifying the genes responsible for this phenotype. RNA isolated from mammary tissues collected at peak lactation (Day 9) from 5 animals in each strain were hybridised to Affymetrix MOE 430 arrays. 134 genes were significantly differentially expressed between the strains with a BY false discovery rate threshold of P < 0.05 and about 973 genes without BY adjustment for multiple testing (P < 0.01). Microarray results were validated by RT-PCR and there was significant correlation between the two methods for all five selected genes. The genes showing differential expression were compared with other lactation and QTL datasets. Previous studies of litter size/ maternal performance which are good indicators of lactation performance have identified QTL on mouse Chr 2, 4 and 7. Integration of the QTL data and expression profiles has identified a number of putative candidate genes for further analysis. One of these candidates, Peg3, a paternally expressed imprinted gene, is overexpressed in the QSi5 strain of mice lies in the QTL region for maternal performance on Chr 7. Peg 3 mutants have impaired lactation and the growth of pups to weaning is consistently lower than wild type dams, similarly, CBA pups have significantly lower growth rate compared to QSi5 strain of mice(p<0.002). 26 SNP polymorphisms are present in this gene in another related inbred strain FVB that has slightly lower lactation performance relative to QSi5 but significantly higher than other inbred strains. Peg3 also lies in Bovine QTL regions and have significant associations for protein traits in recent SNP association analysis. Thus, we have identified Peg 3 as a possible candidate gene responsible for the superior lactation phenotype of QSi5 strain of mice by adopting a comparative genomics approach and this gene is currently the subject of a detailed functional analysis.

WEDNESDAY 15:15 - 15:30 [W5 PHYLOGENY]

### Ancient DNA sheds light on moa gigantism and dwarfism in New Zealand Nic Rawlence<sup>1,\*</sup> Trevor Worthy<sup>1</sup> Jeremy Austin<sup>1</sup> Alan Cooper<sup>1</sup>

<sup>1</sup>University of Adelaide

Keywords: Ancient and Forensic DNA Phylogeny, Biodiversity & Barcoding

Island ecosystems are unique in that they represent natural laboratories where the shackles of evolutionary constraint are loosened allowing the formation of new and varied species. New Zealand is no exception, with moa evolving 10 species ranging in size from giants to species the size of big turkey. Of interest is the propensity for some moa species to exhibit gigantism and dwarfism, which has confounded taxonomy. Recent excavations in the Dart River Valley, South Island have found several dwarfed heavy footed moa (Pachyornis elephantopus) individuals that are potentially a new moa taxa. We use ancient DNA to address the taxonomy of this form. With other gigantism/ dwarfism moa examples we show that even though there are distinct geographic genomic clades within some moa species, they can include giants and dwarfs and do not represent new taxa.

#### WEDNESDAY 15:15 - 15:30 [W6 FUNCTIONAL GENOMICS 1]

### Investigating the target sites of Phenylpyrazole insecticides in Drosophila melanogaster

Emily J Remnant<sup>1,\*</sup> Phillip J Daborn1 Philip Batterham1

<sup>1</sup>Centre for Environmental Stress and Adaptation Research (CESAR), Department of Genetics, Bio21 Molecular and Biotechnology Institute, The University of Melbourne, Victoria, 3010, Australia **Keywords:** Functional Genomics Adaptation

The vast majority of past and currently used insecticides interact with molecular targets involved in neuronal processes. Continued emergence of resistance to neurotoxic insecticides and a necessity to discover more specific insecticide targets has established a need to examine these neural systems in the context of resistance development. An understanding of the molecular interaction of an insecticide with a target site may provide us with information to at best circumvent, or otherwise delay the evolution of resistance mutations.

Fipronil is a phenylpyrazole insecticide widely used in agriculture and animal health. Phenylpyrazoles exert their lethal effects by preventing the actions of inhibitory ligand-gated chloride channels (LGCCs). However questions remain about the specificities of this interaction; in particular, which subunits and residues are crucial for binding? What are the protein-chemical interactions that exist?

The genome sequence of *Drosophila melanogaster* has revealed genes encoding 12 LGCC subunits. Two members of this family, Rdl and GluCla, have been shown to interact with fipronil in previous studies. The potential exists for other non-characterised members of this family to play an important role in toxicity. Two parallel approaches are currently underway to explore phenylpyrazole resistance resulting from LGCC target-site alterations. Ethylmethanesulfonate (EMS) mutagenesis is being used to generate novel mutations resulting in a resistance phenotype. A second approach will utilise recent technologies in *Drosophila* transgenics, incorporating recombineering and site-specific integration to explore mutagenesis of LGCC subunit residues in vivo, in particular Rdl. This approach will be aided by the use of homology modelling of insect receptors and docking studies to predict important residues for binding of the insecticides.

This research will contribute to an understanding of the LGCC neural system in insects, which continues to provide important insecticide targets for present and future insecticides.

#### FRIDAY 14:30 - 14:45 [F5 ADAPTATION]

An adaptive run at an insecticide resistance locus.

Josh Schmidt<sup>1</sup> Rob Trygve Good<sup>1</sup> Emma Warburton<sup>1</sup> Phil Batterham<sup>1,2</sup> Belinda Appleton<sup>1</sup> Charles Robin<sup>1,\*</sup>

<sup>1</sup>University of Melbourne

<sup>2</sup>Centre for Environmental Stress and Adaptation Research **Keywords:** Adaptation Evolutionary Genetics

Insecticide resistance provides a model of adaptation characterized by one of the most intense forms of selection acting on any natural populations of eukaryotes. Because the selection is so extreme, insecticide resistance can inform us about the limits of natural selection in terms of the types of mutations that arise, the frequency at which they occur and re-occur, and about the rate at which adaptive mutations can spread throughout populations. In a remarkable example of parallel evolution, DDT resistant alleles at the Cyp6g1 locus, have recently swept through D.melanogaster and D.simulans populations and in both cases they are defined by the insertion of transposable elements that putatively up-regulate the transcription of this metabolic gene. Here we report that the Cyp6g1 locus from D.melanogaster harbours substantially more allelic diversity than previously appreciated. The locus has been duplicated so that most flies in worldwide populations have more than one copy. Furthermore at least three recent transposable elements have inserted into the 5'regulatory region of these genes. The nested nature of these insertions allows the insertion and duplication events to be placed in a temporal order. Remarkably each new mutation increases the level of DDT resistance it affords. Population samples over temporal and spatial scales suggest that multiple selective sweeps have happened at this locus over the last sixty years. The high frequency of a derived allele in Australia contrasts with its low frequency in contemporaneously sampled North America and Europe populations suggesting this allele may still be sweeping through populations worldwide. The number and magnitude of events that have occurred at Cyp6g1 contrast with contemporary theoretical models of adaptation, which suggest a small number of adaptive steps of decreasing magnitude.

#### FRIDAY 12:45 - 13:00 [F1 POPULATION GENETICS 2]

### **Range Expansions, Rare Mutations And Novel Environments: Evolution In Progress?** Lee Ann Rollins<sup>1,\*</sup> William B Sherwin<sup>1</sup> Andrew P Woolnough<sup>2</sup>

<sup>1</sup>University of New South Wales

<sup>2</sup>Vertebrate Pest Research Section, Department of Agriculture and Food Western Australia **Keywords:** Conservation Genetics Population Genetics Ancient and Forensic DNA

As invasive populations spread across novel environments, opportunities arise for genetic variants normally found in low levels to proliferate. Additionally, variation in selective regimes across different environments may result in local adaptations and allele frequency shifts. Introduced starlings in Australia were sampled across their range, from original introduction sites in the east to western sites on the edge of the range expansion. We investigated whether population-specific DNA sequence polymorphisms in the mitochondrial control region could be identified in samples taken across the introduced range after only approximately 50 generations since introduction. We identified a haplotype found at high frequency which is unique to populations on the edge of the range expansion. Additionally, many individuals carrying this haplotype were heteroplasmic suggesting that this may be a recent mutation. However, further investigation of historical samples identified heteroplasmic individuals from the 19th and 20th Centuries from the native range, indicating that this mutation is persistently heteroplasmic and probably existed at a low frequency in the original introduction. Simulations also indicate that it is realistic to assume that the heteroplasmy could persist since introduction and unlikely that the mutation occurred in Australia. Interestingly, analyses of temporal samples taken on the edge of the range expansion indicate a rapid increase in the ratio of the unique haplotype to the ancestral haplotype over the past four years. Simulations indicate that in order for these observations to be explained by genetic drift alone, population growth rates would need to be extremely high. However, it is possible that selective pressures existing in the novel environment in Western Australia, but absent in Eastern Australia, may also explain the observed genetic trends.

#### WEDNESDAY 12:30 - 12:45 [W2 PHYLOGEOGRAPHY]

## From populations to species: the influence of ecological and environmental factors on rainforest diversity

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<sup>1</sup>National Herbarium of NSW, Botanic Gardens Trust, Mrs Macquaries Rd, Sydney, NSW 2000 **Keywords:** Evolutionary Genetics Speciation and Phylogeography Conservation Genetics Population Genetics

Paleo-ecological, biogeographic and systematic studies provide valuable information on the evolutionary and distributional shifts that shaped the Australian flora. However, as our appreciation of phylogeny and biogeography improves, we are becoming increasingly aware that generalisations about evolutionary dynamics can be untenable without the consideration of ecological and environmental aspects. There are numerous benefits in integrating ecological and genetic approaches to interpret species distribution and population structure. Discrete genetic signatures are left on populations according to how a range of life-history traits can respond to the selective forces operating within current or past environmental processes. Australian rainforest provide an ideal setting for evolutionary studies with high but manageable rates of taxonomic diversity and endemism, and the presence of ancient lineages. Combined genetic and ecological studies are showing that the intricate evolutionary dynamics of rainforest trees is partly due to the tension between persistence and dispersal. An investigation of gene-flow patterns and genetic structure across related and codistributed trees from the genus Elaeocarpus (Elaeocarpaceae) that are distinguishable by their distribution ranges and life-history characters is showing interesting and sometimes unexpected patterns. The combination of molecular and ecological data suggests that the historical influence of recognised biogeographic barriers varies across closely related taxa. This and other studies show that understanding genetic patterns at temporal and geographical scales is only a first step towards the unraveling of evolutionary processes and the development of management strategies. Only by relating these patterns to causal factors we will be able to model the vulnerability of species and ecological communities to environmental change.

FRIDAY 15:15 - 15:30 [F4 CONSERVATION GENETICS 2]

#### Genetic diversity in Murray cod (Maccullochella peelii peelii) across the Murray-Darling Basin.

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#### Keywords: Population Genetics Conservation Genetics

Murray cod are endemic to the Murray-Darling Basin (MDB) of eastern Australia and have considerable cultural and recreational value. Over-fishing, habitat change (such as river regulation) and introduced species have together resulted in a significant decline in population size and distribution. The species is listed under the EPBC act (1999) as vulnerable nationally, indicating it faces a high risk of extinction in the future. Substantial restocking efforts since the early 1980s have used hatchery-raised Murray cod to supplement wild populations for recreational and conservation purposes, with largely unknown effects on genetic structure of wild populations.

We assessed the contemporary genetic population structure of Murray cod over the MDB. Six hundred and eighty contemporary Murray cod from 16 river catchments were screened across 16 polymorphic microsatellites. Preliminary Bayesian analysis of these data revealed that samples from the Lachlan and Macquarie Rivers each were representatives of a discrete genetic cluster. Moderate Fst values (0.05 to 0.13) lend further support to distinction of these catchments. Samples from the Border Rivers, Namoi River and Gwydir River catchments also show some evidence of restricted gene flow, resulting in a slightly lower degree of differentiation than was seen for the Lachlan and Macquarie Rivers. However there is some evidence that the native populations in these catchments may have experienced incomplete introgression with exogenous restocked fish. The remainder of the MDB catchments are apparently one large panmictic population. Thus the genetic data were also revealing about biogeographical processes, namely that Murray cod populations in rivers flowing into the Murray River are highly connected, while those terminating in swamps and wetlands are historically isolated. Temporal comparisons between 200 historical scale samples (up to 50 years old) and contemporary samples from five southern MDB catchments demonstrated limited change in genetic structuring, suggesting little genetic impact of restocking Murray cod in these areas.

Results from this study, when completed, will assist the conservation and management of Murray cod in the wild, as well as provide guidelines for genetically sound breeding and stock enhancement programs.

#### FRIDAY 12:15 - 12:30 [F2 CONSERVATION GENETICS 1]

# An evaluation of the reliability of estimating population abundance with faecal and hair DNA for the endangered species the Spotted-tailed Quoll (Dasyurus maculatus) Monica P Ruibal<sup>1,\*</sup>

<sup>1</sup>Australian National University

Keywords: Conservation Genetics Ancient and Forensic DNA

Despite its potential as a tool for enumerating populations non-invasive DNA sampling has been applied successfully for few mammal species. We assessed the feasibility of estimating population abundance of the endangered species the spotted-tailed quoll (Dasyurus maculatus) using remotely collected faeces and hair from a population in Kosiuszko National Park, Australia. The reliability of the faecal and hair genetic profiles and population estimates were evaluated against genetic and capture data derived by live-trapping. Population estimates were directly comparable as the datasets were equal in temporal and geographical scales and in sampling effort. Faecal sampling and trapping were carried out for 28 days in 2005, and hair tubing and trapping over 17 days in 2006. Microsatellite profiles were derived from 209 facees, 55 hair-traps and trapped individuals. Allelic dropout and false alleles were present in the 10-locus faecal and hair profiles, however using a multiple tubes approach and with comprehensive error checking individuals could be reliably identified. Faecal DNA sampling detected 16 of the 22-trapped individuals and identified three individuals not known from trapping in 2005. Hair-tubing methods detected 11 of the 20-trapped individuals in 2006. Both faecal and hair DNA sampling methods provided unbiased sampling methods detecting males, females, adults and juveniles. Extended faecal and hair tubing sampling periods were able to improve population estimates substantially. Over four months of faecal sampling a further 10 individuals were detected. With three additional periods of hair tubing of 7-9 consecutive days five extra individuals were detected. In conclusion, this study demonstrated that non-invasive DNA sampling offers an unbiased method for sampling individuals and can provide meaningful estimates of population abundance. However when compared with live-trapping efforts a longer sampling period is required to detect equivalent numbers to that detected by trapping. The need for longer sampling periods may limit the use of the noninvasive methods as a stand-alone management tool.

WEDNESDAY 12:30 - 12:45 [W1 COMPARATIVE GENOMICS 2] Natural Killer Complex Genes In Marsupials And Monotremes Claire Elizabeth Sanderson<sup>1,\*</sup> Hannah Veronica Siddle<sup>1</sup> Kathy Belov<sup>1</sup> <sup>1</sup>University of Sydney Keywords: Evolutionary Genetics Comparative Genomics

Natural killer (NK) cells are an integral component of the innate immune system. The receptors are located on the surface of these cells and play an important role in recognising and destroying virally infected cells and tumour cells. Two different regions of the genome encode these receptors, the Leukocyte Receptor Complex (LRC) and the Natural Killer Complex (NKC). The LRC contains genes that encode immunoglobulin-like receptors, whereas the NKC contains genes that encode C-type lectin receptors. We have characterised the NKC genes in two marsupial species, the South American opossum and the tammar wallaby, and one monotreme, the platypus.

One of the defining features of the eutherian NKC is the varying number of Ly49 genes present in each species. Ly49 are present in rodents and horses in high numbers, whereas humans only have one Ly49 pseudogene and other eutherians like cattle, cats, dogs and pigs have a single Ly49 gene (reviewed in Kelley et al., 2005). Preliminary bioinformatic search strategies did not identify Ly49 orthologs in the opossum, tammar wallaby or the platypus genomes. We suggest that Ly49 genes evolved after the divergence of eutherians and marsupials.

The human NKC is located on chromosome 12p13.1 and contains 15 NKC genes, which comprises both killer cell lectin-like receptors (KLRs) and Ctype lectin-like receptors (CLECs). The opossum NKC on chromosome 8 contains 9 NKC genes. Phylogenetic analysis was used to assign these genes to gene families and orthologs of eutherian NKC genes were found. Eutherians and opossums share the same organisation of NKC genes and therefore we infer that the NKC organisation seen in eutherians evolved prior to the separation of eutherians and marsupials. Preliminary searches in the tammar wallaby identified 8 NKC genes. The opossum and the tammar wallaby genomes appear to have a simpler NKC than eutherians. Preliminary searches have identified 35 NKC genes in the platypus genome, suggesting that a gene expansion occurred in the platypus lineage, leading to a highly complex NKC. Implications of these finding will be discussed.

Reference: Kelley J, Walter L, Trowsdale J (2005) Comparative Genomics of Natural Killer Cell Receptor Gene Clusters. PLoS Genet 1(2): e27 doi:10.1371/journal.pgen.0010027

#### WEDNESDAY 11:45 - 12:00 [W1 COMPARATIVE GENOMICS 2]

### Evolution of segmented mitochondrial genomes in the lice of humans and other primates

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<sup>1</sup>Parasitology Section, School of Molecular and Microbial Sciences, The University of Queensland, <sup>2</sup>The Institute for Genomic Research, 9712 Medical Centre Drive, Rockville, MD 20850, U.S.A. **Keywords:** Comparative Genomics Comparative Genomics

We present evidence for the replacement of the typical type of mitochondrial (mt) genome of animals with a group of segments in the human body louse, Pediculus humanus. Unlike most other animals whose 37 mt genes are in a single ~15-kb DNA circle, mt genes of the human body louse are in at least 12 independent segments that are ~3 kb in size. Each segment has 1-3 genes and a large-non-coding region. All of the segments present in the human body louse are also present in the human head lice. Further, one segment that has the gene rmS is also present in the human pubic louse and the langur louse. We found no evidence for the presence of any of these segments in the lice of chimpanzees, gorillas and lutungs. We infer that: (1) some of the mtDNA segments in the human body louse were present in the common ancestor of the lice of humans, other apes and the Old World monkeys, which lived ~22.5 million years ago (MYA); and (2) separated segments replaced the typical type of mt genome in the lineage that led to the human body louse after this lineage and the lineage that led to the chimpanzee louse diverged ~5.6 MYA. Increases in the proportion of separated segments may have been associated with founder events that apparently occurred in the lineage that led to the human body louse, and a selective advantage of these segments in replication over the typical type of mt genome.

#### WEDNESDAY 15:45 - 16:00 [W6 FUNCTIONAL GENOMICS 1]

Challenging strategies in development of cost effective methods from collection and storage of blood samples to high-throughput DNA extraction

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With the advent of rapid developments in genotyping and sequencing technologies, exploration of enormous number of SNPs and also rapid genome sequencing are becoming possible in a variety of species. The 500k SNP chip in human and the 454 sequencing technology are only current examples of such developments. However, easy access to larger numbers of DNA samples to fulfill the requirements of these rapidly growing technologies has not yet well developed. Issues like collection and storage of large numbers of blood samples and extraction of large volumes of DNA are generally associated with time consuming tasks plus attributing considerable amounts to overall genotyping costs. With the current situations it seems that the preparation of large scale DNA samples is a bottleneck for large scale genotyping and needs to be challenged. There are many commercial service providers around the globe that offer DNA extraction services with generally very high costs. In addition and more importantly the starting point for these agencies is to feeding them with already collected and stored samples, while the time consuming and costly task of collection and storage of samples are not mechanized in large scale levels. In this report we provide a comprehensive review of DNA extraction methods and propose strategies for rapid and cost effective methods of large scale DNA extractions.

THURSDAY 11:45 - 12:00 [TH2 EVOLUTIONARY GENETICS]

Instability of chloroplast-derived DNA in the nuclear genome of tobacco Anna Sheppard<sup>1,\*</sup> Michael Ayliffe<sup>2</sup> Jeremy Timmis<sup>1</sup> <sup>1</sup>University of Adelaide <sup>2</sup>CSIRO Plant Industry Keywords: Evolutionary Genetics Plant Genetics

DNA transfer from chloroplast and mitochondrial genomes to the nuclear genome is a frequently occurring process in eukaryotes. To investigate the nature of this transfer, a kanamycin resistance gene (nptII) designed for exclusive nuclear expression was inserted into the chloroplast genome of tobacco. Progeny from backcrosses were grown on medium containing kanamycin to select for nuclear transposition of nptII and a number of independent hemizygous plants were obtained, demonstrating a transfer frequency of approximately 1 event per 16,000 pollen grains.

In contrast to Mendelian expectations, for some of these plants significantly less than 75% of progeny resulting from self fertilisation were kanamycin resistant. For 9 independent integrants, the nature of this instability has been characterised in detail by collecting many individual self fertilised seed capsules and determining the percentage of kanamycin resistant progeny for each capsule. Of these, 4 integrants gave the expected 75% kanamycin resistant progeny in all seed capsules, 1 gave approximately 50% kanamycin resistant progeny in all seed capsules and the remaining 4 showed variation between seed capsules, with levels of kanamycin resistance ranging from 0% to 75%. The molecular basis of this instability has also been investigated and PCR analysis indicates that deletion of nptII is likely to be the mechanism responsible. Given the high transfer frequency of DNA from the chloroplast genome to the nuclear genome, it would be expected that the size of the nuclear genome would increase significantly over a relatively short evolutionary time, unless there is a counterbalancing removal of some integrants. The work described here provides evidence that ingress and egress of organellar DNA to and from the nucleus are in dynamic equilibrium.

#### WEDNESDAY 12:45 - 13:00 [W1 COMPARATIVE GENOMICS 2]

### Class I MHC genes in the tammar wallaby (Macropus eugenii) are dispersed throughout the genome Hannah V Siddle<sup>1,\*</sup> Janine E Deakin<sup>2</sup> Penny Coggill<sup>3</sup> Stephan Beck<sup>3</sup> Katherine Belov<sup>1</sup>

Hannah V Siddle'' Janine E Deakin' Penny Coggill' Stephan Beck' Katherine Belov' <sup>1</sup>University of Sydney <sup>2</sup>ARC Centre for Kangaroo Genomics, Australian National University <sup>3</sup>Wellcome Trust Sanger Institute

Keywords: Comparative Genomics Evolutionary Genetics Functional Genomics

The major histocompatibility complex is a large multigene family, responsible for antigen recognition in all vertebrates. MHC genes are classified into three groups, Class I, Class II and Class III. In eutherian mammals the three groups are genetically linked, which is thought to be important for the coevolution of these genes. Class I genes evolve rapidly through gene duplication and the number of Class I loci can vary greatly between different species. Some Class I genes are not involved in antigen presentation and have taken on more divergent functions, such as foetal-maternal tolerance, these genes are known as non-classical Class I.

We have isolated tammar wallaby BACs containing Class I, II and III genes. The Class II and III genes map to chromosome 2, while the Class I genes are dispersed throughout the genome on six different chromosomes. This remarkable MHC organisation has not been observed in any other mammalian species. We have sequenced 10 entire BACs containing Class I genes from 6 different chromosomes, in order to elucidate the evolutionary history of the Class I dispersal. Sequence analysis has allowed us to identify classical and non-classical genes based on coding sequences and promoter regions. Our results raise questions about the importance of clustering of immune genes.

### TUESDAY 18:40 - 18:55 [S1 STUDENT]

#### Devil Facial Tumour Disease: Lack of histocompatibility barriers explains the spread of a transmissible tumour

Hannah V Siddle<sup>1,\*</sup> Mark DB Eldridge<sup>2</sup> Erin Noonan<sup>3</sup> Stephen Pyecroft<sup>3</sup> Katherine Belov<sup>1</sup> <sup>1</sup>University of Sydney <sup>2</sup>Australian Museum

<sup>3</sup>Animal Health Laboratory, Department of Primary Industries and Water

Keywords: Conservation Genetics Population Genetics Comparative Genomics

The largest remaining marsupial carnivore, the Tasmanian devil, is under threat of extinction from a transmissible tumour, Devil Facial Tumour Disease (DFTD). DFTD is thought to be a rogue cell line, spread between devils by biting around the face and neck. Spread of the tumour should be prevented by highly polymorphic, major histocompatibility complex (MHC) molecules that recognise foreign grafts. However, recognition and immune response to the tumour does not occur. There are two possible genetic causes for the spread of DFTD. First, the tumour may alter MHC gene expression to evade the host immune system. Second, the devil population may lack diversity at MHC loci, preventing recognition of the tumour as foreign.

Here we show that devils have remarkably low diversity at MHC loci, resulting in failure of the devil immune system to mount a response to the tumour. We demonstrate that DFTD cells express a range of functional MHC genes and conclude that altered MHC expression is not responsible for spread of the tumour. Further, we provide genetic evidence that DFTD is a clonal cell line by genotyping matched tumour and host samples. The Tasmanian devil has undergone a 'bottleneck' at MHC loci, perhaps during population crashes recorded in the past 150 years. Low diversity at functionally important loci, such as the MHC, should be considered when planning captive breeding programs for devils. Further, lack of histocompatibility barriers in the devil population means the only way to control the disease is to remove affected individuals from the population.

WEDNESDAY 11:45 - 12:00 [W2 PHYLOGEOGRAPHY]

### Phylogeography of the Amazonian Pencilfish Nannostomus unifasciatus (Lebiasinidae) Using Intron DNA Markers

Mark J Sistrom<sup>1,\*</sup> Ning L Chao<sup>2</sup> Luciano B Beheregaray<sup>1</sup> Macquarie University <sup>2</sup>Department of Fisheries, Universidade do Amazonas

Keywords: Speciation and Phylogeography Conservation Genetics

The Amazon basin is home to the highest diversity of freshwater fishes on Earth. However, the evolutionary mechanisms responsible for the development of this diversity are poorly understood. The pencilfish Nannostomus unifasciatus (Lebiasinidae) is a small forest fish found in association with flooded forest environments of the Rio Negro Floodplain (RNF), in central Amazonia. This species is harvested commercially in the RNF aquarium fishery, and as such contributes to the livelihood of riverine communities of this remote region. We amassed a large sample of N. unifasciatus (n = 247) from 20 tributaries of the Rio Negro, an effort that effectively covers the entire distribution of the species in this river basin. Here we present the results of a large scale phylogeographic analysis of N. unifasciatus based on sequence data from the second intron of the S7 ribosomal protein. Two highly divergent lineages with complex geographic distributions were detected. Overall, population differentiation was markedly stronger in the headwater region. Conversely, intra-population genetic diversity was lower in the headwaters and higher downstream. Although these results are generally consistent with previously collected mitochondrial DNA and microsatellite data, the more ancient

temporal scale provided by the intron-based analysis has substantially increased our understanding of N. unifasciatus phylogeography. This study provides critical information for the conservation management of the N. unifasciatus ornamental fishery and validates the usefulness of the second intron of the S7 ribosomal protein as a marker for phylogeographic studies.

#### WEDNESDAY 11:00 - 11:15 [W2 PHYLOGEOGRAPHY]

## Unexpectedly ancient cryptic species and global oceanic dispersal in the smallest eukaryotes

Jan Slapeta<sup>1,\*</sup> Purification Lopez-Garcia<sup>2</sup> David Moreira<sup>2</sup> <sup>1</sup>Faculty of Veterinary Science, University of Sydney, Sydney, Australia <sup>2</sup>Unite d'Ecologie, Systematique et Evolution, Universite Paris-Sud, Orsay, France Keywords: Speciation and Phylogeography Environmental Microbes

Small eukaryotes (smaller than 1 mm) are thought to behave as prokaryotes in that, lacking geographical barriers to their dispersal due to their tiny size, they are ubiquitous and represented by a relatively small number of species. Accordingly, the absence of geographical isolation would imply the existence of a relatively small number of microeukaryotic species. To test these ideas, we sequenced and compared several nuclear, mitochondrial and chloroplast genes from isolates of a marine picoeukaryotic alga (approximately 2 micrometers), Micromonas pusilla, collected worldwide. Independent and combined phylogenetic analyses demonstrate that this traditional single morphospecies actually comprises several independent lineages, some of which are shown to be ubiquitous in oceans. However, while some lineages group closely related strains, others form distant clusters, revealing the existence of cryptic species. Moreover, molecular dating using a relaxed clock suggests that their first diversification may have started as early as during the Late Cretaceous (approximately 65 million years ago), implying that M. pusilla is the oldest group of cryptic species known to date. Our results illustrate that global dispersal of a picoeukaryote is possible in oceans, but this does not imply a reduced species number. On the contrary, we show that the morphospecies concept is untenable since it overlooks a large genetic and species diversity, and may lead to incorrect biological assumptions.

### WEDNESDAY 12:45 - 13:00 [W3 BIOINFORMATICS]

# Comparison of false discovery rate controlling procedures for microarrays Jie $Song^{1,2,*}$ Peter Thomson<sup>1,2</sup>

<sup>1</sup>Faculty of Veterinary Science, University of Sydney <sup>2</sup>CRC for Innovative Dairy Products

Keywords: Bioinformatics Functional Genomics

In the microarray context, False Discovery Rate (FDR) is the proportion of genes which are declared to be differentially expressed (DE) but which in fact are not. The FDR controlling procedures can be used to identify DE genes for microarray experiments. Several FDR controlling procedures including four presented by Reiner et al. and the q-value method proposed by Storey and Tibshirani (ST) have been proposed. In this study, we modified these FDR methods, and used simulation based on the mixture model which includes two populations, DE genes and non-DE genes, to evaluate the performance of these five FDR controlling procedures for one-sample microarray studies. We simulated a wide range of scenarios. The first simulation scenario assumed a common noise (error) variance for each gene and gene independence. Then we simulated increasingly complex scenarios where the error variance was not constant (drawn from a gamma distribution) and gene dependence facilitated through introducing gene clusters. Finally, it is found that the ST point-estimate FDR procedure is the best procedure for estimating FDR for most simulation scenarios based on the mean square error (MSE) criteria. The MSE of the best procedure is the smallest one among all the procedures for each scenario. We applied ST point-estimate FDR controlling procedure to analyse one real public one-sample microarray data set (CD4+ T cell data set) which consists of 27 arrays and 21,380 genes, and found 1768 differentially expressed genes using a FDR of 5%, or equivalently ~ 88 false discoveries.

FRIDAY 12:15 - 12:30 [F1 POPULATION GENETICS 2]

Using linkage disequilibrium to estimate effective separation times for different worldwide human populations.

John Sved<sup>1,\*</sup> Allan McRae<sup>2</sup> Peter Visscher<sup>2</sup>

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Keywords: Population Genetics Bioinformatics

We have used the Hapmap database to calculate values of riri for different pairs of populations, where riand  $r_j$  are correlations of alleles for a pair of loci in populations *i* and *j*. If there is a level of linkage disequilibrium  $r^2$  0 in the parent population before separation, it is expected that, for loci separated by recombination fraction c, rirj will be reduced from this value by an amount  $(1-c)^2$  in each generation. The advantage of such a two-locus calculation over traditional single locus FST calculations is that population size should not affect the estimate of divergence. - Over 250,000,000 pairs of loci (SNPs) within 1 cM are available for the linkage disequilibrium calculation. Cumulated over all such pairs, we find that there is a positive signal in r values up to 0.3 cMs for the separation between African and non-African populations. One potential problem in our approach is that we require linkage disequilibrium estimates in the ancestral parent population (r2 0), whereas we observe linkage disequilibrium in present day populations. In addition, the  $(1-c)^2$  expectation is sensitive to two forces, one of which we call fixation bias which is particularly important for very low recombination frequencies, and the other, recombination frequency mis-estimation, which is potentially important for higher recombination frequencies. Taking all this into account, we estimate the effective separation time of African and non-African populations as less than 1,000 generations. Such a time can only be consistent with current estimates of historical separation if there has been substantial migration between African and non-African populations.

#### THURSDAY 11:15 - 11:30 [TH2 EVOLUTIONARY GENETICS]

#### Two independent duplications forming the Cyp307a genes in Drosophila.

Tamar Sztal<sup>1,\*</sup> Henry Chung<sup>1</sup> Lydia Gramzow<sup>1</sup> Phillip J. Daborn<sup>1</sup> Philip Batterham<sup>1</sup> Charles Robin<sup>1</sup> <sup>1</sup>Centre for Environmental Stress and Adaptation Research (CESAR), Department of Genetics, Bio21 Molecular and Biotechnology Institute, The University of Melbourne, Victoria, 3010, Australia **Keywords:** Comparative Genomics Comparative Genomics

Molting, an essential process required for insect growth, is modulated by a number of key developmental hormones. 20-hydroxyecdysone (20E) is a major molting hormone and is responsible for coordinating many of the physiological and biochemical processes during molting and insect development. A number of cytochrome P450 genes, termed Halloween genes, encode enzymes responsible for the synthesis of 20E from dietary cholesterol precursors. Both the expression patterns and biochemical functions of these Drosophila genes and their orthologues in many insect species are conserved. We present evidence that the Drosophila Cyp307a genes, essential in the biosynthesis of 20E, do not follow the same conserved evolutionary pattern. Using phylogenetic analyses we show that the Drosophila Cyp307a1 (spook) and Cyp307a2 (spookier) genes were formed from two independent duplication events within the Drosophila genus, depicting a complicated evolutionary scenario. An initial duplication, from a Cyp307a2 ancestral gene produced the Cyp307a1 gene, maintained only in the Sophophoran subgenus. A second duplication in the Drosophila subgenus only, formed an additional paralog, Cyp307a3. Microsynteny is conserved for Cyp307a2 throughout the Drosophila species, but not for Cyp307a1 or Cyp307a3. These are located in different genomic positions in the Sophophora and Drosophila subgenera respectively. Cyp307a3 appears to encode a functional gene product and is expressed in a different spatial and temporal manner to Cyp307a1. This suggests some level of functional divergence between the Cyp307a paralogs in different Drosophila species.

#### THURSDAY 11:15 - 11:30 [TH1 POPULATION GENETICS 1]

#### Molecular characterisation of Microsatellite DNA and EPIC DNA markers of the pest moth Helicoverpa armigera

Wee Tek Tay<sup>1,\*</sup> Gajanan T Behere<sup>1</sup> Siu Fai Lee<sup>1</sup> David G Heckel<sup>2</sup> Phil Batterham<sup>1</sup>

<sup>1</sup>Department of Genetics, Bio21 Molecular Science and Biotechnology Institute, The University of Melbourne, Victoria 3010, Australia.

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**Keywords:** Population Genetics Evolutionary Genetics

Understanding the molecular ecology and population structure of an organism requires reliable and robust nuclear DNA markers. Over the past two decades, microsatellite DNA markers have been the markers of choice in evolutionary and population genetic studies, although their application in understanding lepidopteran population genetic structures has been limited. Developing microsatellite DNA markers in Lepidoptera is widely acknowledged as challenging, and this applies also to the Old World polyphagous bollworm Helicoverpa armigera. The few population studies based on microsatellite DNA markers developed for H. armigera have encountered problems such as allele "drop out", excess of homozygosity, and the presence of microsatellite DNA families leading to multiple alleles being detected within individuals. Furthermore, due to the wide geographic distribution of H.

armigera, microsatellite DNA markers developed from individuals that originated from specific populations typically have limited success when applied to other populations. In this study, we characterised in detail H. armigera microsatellite DNA alleles to understand the molecular basis underlying these problems. As an alternative, we propose the use of Exon-Primed Intron-Crossing (EPIC) DNA markers. The applicability of published microsatellite DNA markers and new H. armigera EPIC markers in other Old and New World Helicoverpa pest species will also be compared and discussed.

#### WEDNESDAY 14:00 - 14:15 [W5 PHYLOGENY]

## Molecular markers for understanding the evolutionary and population genetics of the pest moth genus Helicoverpa

Gajanan T Beherel Wee Tek Tay<sup>1,\*</sup> Derek A Russell<sup>2</sup> David G Heckel<sup>3</sup> Phil Batterham<sup>1</sup> <sup>1</sup>Dept Genetics, Bio21 Molecular Science and Biotechnology Institute, University of Melbourne <sup>2</sup>Natural Resources Institute, University of Greenwich, Chatham Maritime, Kent, Me4 4TB, UK <sup>3</sup>Max-Planck Institute for Chemical Ecology, Department of Entomology, Hans-Knöll-Straße 8, D-07745 Jena, Germany.

Keywords: Phylogeny, Biodiversity & Barcoding Evolutionary Genetics

The genus Helicoverpa includes some of the most devastating agricultural lepidopteran pest species of the Old and New Worlds. Within the Old World, H. armigera is a polyphagous species that feeds on over 150 different host crops in Africa, Asia, Europe and Australia. The oligophagous H. assulta that feeds only on Solanaceous crops is found in Africa, Asia and Australia, and the polyphagous H. punctigera is endemic to Australia. In the New World, H. zea has attained a similar pest status as that of H. armigera. Despite the importance of the genus Helicoverpa, there has been limited knowledge on the molecular genetic evolutionary relationships among these four, morphologically similar pest species. Accurate and rapid identification of these species is especially important when taking into consideration international movements of agricultural commodities. Previously, Helicoverpa phylogenies were inferred from nuclear genes based on limited samples, while no mitochondrial DNA (mtDNA) phylogenies have been inferred. Here, we present a mtDNA COI partial gene phylogeny for the four pest Helicoverpa species, and show that H. armigera sampled widely from Australia, Africa and Asia represent a single species. H. zea from both North and South America also represent a single species, and share close evolutionary relationships with H. armigera, suggesting possible founder events from H. armigera at approximately 1.5 million years ago. MtDNA PCR-RFLP analyses indicates that robust and rapid species identification can be easily achieved based on COI and Cyt b genes at all life stages, thereby providing an important identification tool for quarantine inspections of exotic pests, and aiding management strategies due to the differential response to insecticides in these four Helicoverpa species.

#### WEDNESDAY 11:00 - 11:15 [W1 COMPARATIVE GENOMICS 2]

integrated understanding of microbial ecology.

#### Genomics and metagenomics of marine and Antarctic microorganisms

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<sup>2</sup>School of Biotechnology and Biomolecular Sciences, The University of New South Wales **Keywords:** Environmental Microbes Comparative Genomics

Microorganisms are key players in all environmental systems and perform functions ranging from biogeochemical cycling and primary energy production to pathogenicity of higher organisms. Despite this level of understanding it is clear that more than 99% of the natural, microbial diversity stills remains to be explored. A recent focus on genome sequencing of environmental microorganisms has ushered in a new phase of understanding their genetic and physiological properties. Major technological advances have recently propelled the field forward by instigating programs of DNA sequencing of whole environmental samples of microorganisms (metagenomics). In this presentation we will overview findings from comparative genomics of several marine and Antarctic model microorganisms. Examples will be given that demonstrate how genomics has facilitated our understanding of the production of bioactive compounds, prokaryote/ eukaryote interactions, gene regulation and thermal adaptation in these organisms. We will also discuss two metagenome projects for marine surface and Antarctic ecosystems and will describe how meta/genome-based studies can not only provide insight into microbial diversity, but particularly when linked to functional studies (e.g. proteomics, functional screens) can deliver a comprehensive view of microbial adaptation and an

#### TUESDAY 19:10 - 19:25 [S1 STUDENT]

## Impact of Heavy-Metal Pollution on Sediment Microbial Community Diversity in an Urban Creek

Carly P Tucker<sup>1,\*</sup> Vincent Pettigrove<sup>2</sup> Hatch W Stokes<sup>3</sup> Yvonne M Parsons<sup>1</sup>

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Keywords: Environmental Microbes Adaptation

Anthropogenic disturbances represent a significant threat to the health of river ecosystems. The aim was to investigate the impact of long-term heavy metal pollution on the microbial community composition in Merri Creek, Victoria. Sediment was collected from 4 distinct locations: an upstream reference site, a highly polluted site and two moderately polluted sites. Sediment bacterial 16S rRNA transcripts were amplified using RT-PCR and approximately 100 clones per site were sequenced from the 5' end. Sequence classification using the RDP database (http://rdp.cme.msu.edu/) showed that the majority of clones from the most polluted site were readily identifiable, while a significant proportion of clones from the reference site could not be identified with certainty, even at the class level. Additionally, the classification suggested that the lowest number of classes was found at the most polluted site and that the highest number was found at the reference site. This was further investigated using DOTUR (Distance-Based OTU and Richness Determination), a program that analyses distance matrices and assigns sequences to operational taxonomic units (OTU) for all possible genetic distances before calculating curves for sampling intensity, richness estimators and diversity indices (http://www.plantpath.wisc.edu/fac/joh/dotur.html). Construction of lineage through time plots shows that the extinction of phylogenetic lineages occurs faster than the creation of new ones at the polluted sites, and that this rate is fastest at the most polluted site. Comparison of rarefaction curves constructed with OTUs defined at 3% and 10% genetic distance show that the reference site and the most polluted site are significantly different at each level, but they could only be separated from the moderately polluted sites at the 10% level. This study shows that differences in sediment microbial community diversity can be correlated with heavy metal pollution, especially when the distance used to define an OTU is increased

THURSDAY 12:30 - 12:45 [TH2 EVOLUTIONARY GENETICS] Cane toad (Bufo marinus) toxin resistance in goannas Beata Ujvari<sup>1,\*</sup> Thomas Madsen<sup>1</sup> <sup>1</sup>School of Biological Sciences, University of Wollongong Keywords: Evolutionary Genetics Population Genetics

The recent invasion of cane toads into the Northern Territory of Australia has resulted in a massive increase in mortality of some native squamate predators such as goannas. However, our field work demonstrate that a small number of goannas are able to persist in cane toad infested areas. Recent studies have suggested that propensity to feed on toxic prey, and/or morphological change could explain why some squamate predators are able to persist in such areas. We have recently identified a gene that provides resistance to toad toxins in some squamate reptiles, and therefore suggest that an additional explanation for the persistence of goannas in cane toad infested areas could involve selection on cane toad toxin resistance. As no members of the family Bufonidae are native to Australia, we propose that such selection should be traced to the evolutionary history of Australian goannas. These carnivorous lizards did not originate in Australia, and arrived to the Australian continent about 10 million years ago. The diet of all non-Australian goannas frequently include toads, demonstrating that all of these taxa are resistant to toad toxins, and strongly suggests that when arriving to the Australian continent, goannas were resistant to toad toxins. However, the absence of toad on the Australian continent most likely resulted in a relaxed selection in maintaining such a resistance. Our preliminary studies demonstrate that non-Australian goannas harbour the resistance gene, whereas Australian goannas do not. However, we have documented a substantial individual variation in this gene in the indigenous, Varanus panoptes, albeit none, so far, having an exact copy of the resistance gene. If a gene providing increased resistance to cane toad toxin is the main selective agent ensuring survival, we expect that the frequency of such a gene would increase dramatically among the surviving goannas.

#### THURSDAY 12:00 - 12:15 [TH1 POPULATION GENETICS 1] The genetic and behavioural plasticity of montane populations Kate Umbers<sup>1,\*</sup> <sup>1</sup>Macquarie University Keywords: Conservation Genetics Population Genetics

Variation in genetics and behavioural strategies between species is common and well reported, however, variation and plasticity within species has received very little attention. Plasticity in mating strategy and the ability to switch genes on and off allows individuals of a species to gain maximum fitness benefit within dynamic environmental constraints. Individuals within populations whose habitat spans altitudinal gradients for example, may exhibit different strategies at high elevation compared to low. This project will quantify variation and plasticity in various animal populations occurring across altitudinal gradients in the Australian alpine region. I predict that the apparent environmental differences, in particular temperature and UV exposure, at varying altitudes will be reflected in protein production the outcomes of reproductive events in these populations. Grasshoppers of the genus Kosciuscola and are of interest as their reproductive output and body colour are known to vary with altitude. Skinks of the genus Pseudemoia and cockroaches of the genus Polyzosteria will also be targeted as they are highly abundant and widely distributed in the alpine region. Ectothermic animals are of particular interest due their physiological requirements and consequent behavioural adaptations to environmental temperature and other variables. Paternity studies of clutches (using microsatellites and AFLP) will assess genetic mating systems while data on clutch number and size, offspring sex ratio, and timing of reproduction will address behavioural plasticity. Field transplant experiments and common garden treatments will further enhance the scope of this project.

This is the subject of my PhD research which I began this year - feedback is welcomed!

#### WEDNESDAY 12:15 - 12:30 [W3 BIOINFORMATICS]

## A grid of Oxford grids: a comparative genomics tool for Ensembl's automatically annotated genomes

Jonathan M. Usmar<sup>1,2,\*</sup> Matthew Hobbs<sup>1,3</sup> Frank W. Nicholas<sup>1,2,3</sup>

<sup>1</sup>Reprogen, Faculty of Veterinary Science, University of Sydney, NSW 2006

<sup>2</sup>SheepGenomics, Faculty of Veterinary Science, University of Sydney, NSW 2006

<sup>3</sup>CRC for Innovative Dairy Products, Faculty of Veterinary Science, University of Sydney, NSW 2006 Keywords: Comparative Genomics Bioinformatics

An Oxford grid comprises a square divided into a number of rectangles, each representing two chromosomes (one from each of two species), with the length of the sides proportional to the length of those two chromosomes. Each orthologue common to the two species is represented by a point indicating its location on the relevant chromosome of both species. Ensembl (http://www.ensembl.org) is a project for automatic annotation of sequenced genomes. The current Ensembl data release (v44) comprises 34 species. Ensembl data includes comparative genomics data produced by BlastZ-net, translated blat, MLAGAN and gene homology analysis, and a part of this is the automatic production of lists of orthologous and paraloguous Ensembl genes. Information about orthologues is provided by Ensembl in a format that is easy to retrieve and use. We have developed a tool that automatically generates a zoomable Oxford grid of Ensembl orthologues for each pair-wise combination of the species in Ensembl, thereby providing a visualization of the comparative map of each pair of species. The grids are automatically updated with each new release of the Ensembl database. The resultant grid of Oxford grids is available at <u>http://oxgrid.angis.org.au/oxg\_table.html</u>.

WEDNESDAY 14:45 - 15:00 [W4 COMPARATIVE GENOMICS 3]

#### Opossum X Chromosome Sequence Reveals Steps In The Evolution Of The Human X And X Inactivation

Matthew J Wakefield<sup>1,2,\*</sup> Tarjei S Mikkelsen<sup>3</sup> Andrew J Gentles<sup>4</sup> Jennifer AM Graves<sup>5</sup> John M Greally<sup>6</sup> The Opossum Genome Sequencing Consortium<sup>3</sup>

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<sup>2</sup>The ARC Centre for Kangaroo Genomics

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<sup>4</sup>Radiology Department, Stanford University and Genetic Information Research Institute, CA, USA

<sup>5</sup>ARC Centre for Kangaroo Genomics, Research School of Biological Sciences, Australian National University, Canberra, ACT, Australia.

<sup>6</sup>Albert Einstein College of Medicine, Bronx, NY, USA.

Keywords: Comparative Genomics Epigenetics Bioinformatics Evolutionary Genetics Functional Genomics RNAi and non-coding DNA

Sequence analysis of the X chromosome from the marsupial Monodelphis domestica defines the ancient region of the human X, and many marsupialspecific inversions. An XIST gene is absent and accumulation of L1 long interspersed nuclear elements (L1 LINEs) on the X chromosome is significantly less than in human.

These observations support the theory that the X inactivation centre and XIST evolved in the eutherian ancestor only 180-100MYA, modifying a preexisting paternal X inactivation system.

The presence of the XIST gene in eutherian mammals correlates with fewer inversions and increased accumulation of L1 elements, suggesting selection against rearrangements that disrupt the spread of X inactivation from a centre, and exaptation of a pre-existing repeat distribution into the eutherian X inactivation pathway.

Mikkelsen et al (2007) Genome of the marsupial Monodelphis domestica reveals innovation in noncoding sequences. Nature 477:167-77.

Hore et al (2007) XIST is absent from the X chromosome, and its flanking region is disrupted in nonplacental mammals. Chromosome Research 15:147-61

#### FRIDAY 11:45 - 12:00 [F3 MAPPING]

**Map Integration And Genome Comparison Involving The Tammar Wallaby** Chenwei Wang<sup>1,2,3,\*</sup> Kyall Richard Zenger<sup>1,2,3</sup> Lee Webley<sup>1,3</sup> Amber Alsop<sup>3,4</sup> Frank W. Nicholas<sup>1,2</sup> Matthew Hobbs<sup>1,2</sup> Jenny Graves<sup>3,4</sup>

<sup>1</sup>ReproGen, Faculty of Veterinary Science, University of Sydney, NSW, Australia

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<sup>4</sup>Comparative Genomics Group, Research School of Biological Sciences, Australian National University, Canberra, Australia

Keywords: Comparative Genomics Bioinformatics

Marsupials are of significant importance in comparative genomics, given their evolutionary midpoint position in the vertebrate evolutionary tree. The tammar wallaby (Macropus eugenii) is the model marsupial species in Australia. Despite its obvious importance, the genetic architecture and genomic resources of this species are quite limited.

This study aims to build comprehensive integrated genetic maps and analysis tools for the tammar wallaby. This is being carried out via constructing a denser linkage map, integrating the linkage map with physical mapping and sequence data, and developing wallaby genome analysis tools for comparison with other species.

In order to extend the original wallaby linkage map (comprising 64 loci), 76 new microsatellite markers have been identified from physically located or end-sequenced BAC clones. To-date, 48 of these markers have been added to the linkage map using 353 informative meioses from hybrid phaseknown backcross tammar wallabies, resulting in a linkage map comprising 102 loci. This linkage mapping data has been integrated with physical mapping data by using 42 loci common to both maps. This enabled all 9 linkage groups to be assigned to chromosomes. All available mapping information has been incorporated into a single integrated map comprising a total of 377 loci, by applying Location DataBase (LDB) program.

Based on the tammar wallaby integrated map, a draft comparative map between human and tammar wallaby has been created which contains 210 orthologues. Additional orthologous loci are being sought by mining and comparing the tammar wallaby genome sequence with other species, in particular the opossum. OxfordGrids are employed as a means of visualising comparative maps. Through the study of breakpoints in comparative maps with sequences species such as human and opossum, a virtual tammar wallaby genome is within our reach.

WEDNESDAY 15:15 - 15:30 [W4 COMPARATIVE GENOMICS 3]

Which came first: Mammals or the X and Y? Paul D Waters<sup>1,\*</sup> Jennifer A.M Graves<sup>1</sup>

<sup>1</sup>Comparative Genomics Group, Research School of Biological Sciences, The Australian National University, Canberra, ACT, Australia

Keywords: Comparative Genomics Evolutionary Genetics

In most mammals females have two X chromosomes, whereas males have only one X and a small male determining Y that evolved from an ancestral X via a process of degradation and gene loss. An exception to this is the platypus (a monotreme mammal), which has 5 Xs and 5 Ys that form a chain at meiosis. One end of this chain (X5) contains genes orthologous to the chicken Z, whereas at the other end (X1) it was thought that there were genes homologous to the human X. These observations led to the hypothesis that monotreme sex chromosomes might represent an intermediate evolutionary step between a chicken like ZZ/ZW sex chromosome system and a human like XX/XY system. However, recent mapping of regions orthologous to the human X in platypus has demonstrated that it shares no homology with the X1. This finding has widespread implications for the timing of events in mammalian sex chromosome evolution. The monotreme meiotic chain does not represent a link between the bird ZW and mammal XY systems; rather it arose in monotremes from the ZW after their divergence from other mammalian radiation but before marsupial and placental mammals split 180MYA and, therefore, are much younger than previously thought.

### TUESDAY 12:15 - 12:30 [T1 COMPARATIVE GENOMICS 1]

## The origin of platypus venom molecules

Camilla Whittington<sup>1,\*</sup> Anthony Papenfuss<sup>2</sup> Katherine Belov<sup>1</sup> <sup>1</sup>Faculty of Veterinary Science, University of Sydney, NSW <sup>2</sup>The Walter and Eliza Hall Institute of Medical Research, Parkville, Vic Keywords: Evolutionary Genetics

Platypuses and other monotremes occupy an important position as an early offshoot from the evolutionary lineage leading from reptiles to mammals. This means that research into the platypus genome offers a unique opportunity for the study of comparative genomics. It is also a chance to unravel the evolutionary history of one of the most interesting features of the platypus: its venom.

Male platypuses possess spurs that are used as offensive and defensive weapons to deliver a complex mixture of venom peptides into the victim. Platypus envenomation can kill dogs, and is reported to be excruciatingly painful in humans, yet our knowledge of platypus venom is incomplete. It is known that the venom contains peptide fractions like defensin-like peptides (vDLPs), but there are many more unidentified fractions. Considering the range of useful molecules that have been identified in snake venom, it is hoped that research into the pharmacology of platypus venom may yield novel drugs and new targets for painkillers.

Working as part of the platypus genome sequence annotation team, we have characterised some of the venom genes of the platypus. The genome of the platypus was mined for the gene sequences of molecules sharing homology with the partial venom peptide sequences previously obtained by mass spectroscopy (Torres et al., 1999). Identification of signals, such as conserved protein structures, gene duplications and clustering of venom genes on particular chromosomes has provided us with clues as to how these genes have arisen. Interestingly, it appears that they have evolved through duplication of existing immune genes. This research has paved the way for further research into mammalian venom, and provides an intriguing glimpse into mammals' evolutionary past.

#### FRIDAY 14:45 - 15:00 [F5 ADAPTATION]

### **Enigma of the yellow mutation of the cotton bollworm Helicoverpa armigera** Adam Williams<sup>1,2,\*</sup> Philip Batterham<sup>1</sup> David Heckel<sup>2</sup>

<sup>1</sup>Bio21, University of Melbourne, <sup>2</sup>Max Planck Institute for Chemical Ecology, Jena, Germany **Keywords:** Comparative Genomics Adaptation

The yellow mutant of Helicoverpa armigera has depigmented eggs, white to green larvae, white larval stemmata and yellow adult eyes. In an interaction with another mutant, dark, an even more depigmented eye phenotype termed zombie is generated. The zombie line is a potentially valuable strain for screening transgenic insects using the 3xP3 eye enhancer as a driver, or complementation with a wildtype yellow vector. The latter, is only possible if the affected gene is identified. Cloning the promoter would also provide a native enhancer active at all life stages, another valuable transgenesis tool. We have systematically tested two gene pathways and eliminated four genes as the causative lesion in yellow. The ommochrome synthesis pathway is disrupted in vermilion and cinnabar mutants in several insect orders, causing a depigmented eye. Both mutants are rescued by administration of 3hydroxy-kynurenine in larval diet. Experiments on yellow larvae versus a cinnabar mutant (Drosophila Celera) failed to complement yellow, while complementing the cinnabar mutant dose dependently. Moreover yellow does not display maternal inheritance rescue of egg pigment as seen in the Bombyx silkworm cinnabar mutant. The second pathway involves the ABC transporters white and scarlet that pump ommochromes into pigment granules in retinal cells. The full-length transcript of white and most of the 5' of scarlet was cloned in armigera by degenerate PCR and RACE. Both have very high homology to the Drosophila genes and are virtually identical to their Bombyx homologues. In the process the full length Bombyx homologue of scarlet was also identified and cloned for the first time. A variety of molecular mapping techniques on a yellow segregating backcross family revealed that neither gene was linked to the yellow chromosome, however they are themselves linked. There were no errors or consistently segregating SNPs in the yellow cognate genes and RT-PCR levels were generally normal. ABCs are a divergent superfamily with over 30 in Bombyx, so a novel ABC may be the causative gene. RNAi against the white gene using siRNA produced a phenocopy of the yellow mutant at egg and first instar stages, with ~90% phenotypic and mRNA knockdown observed. Experiments to knockdown larval and adult eye pigment are ongoing. 3 kb upstream of the white transcription start site has been amplified, most sequenced, and should prove useful as a native transgenic promoter. Positional cloning of yellow is now underway using a high throughput AFLP approach on a large mapping family.

#### FRIDAY 12:15 - 12:30 [F3 MAPPING]

## A Comaprative Study Of The Abc-Family Transporter, ABCG2, As A Lactation Associated Candidate Gene

Jerry CY Weil Julie A Cavanaghl Paul A Sheehyl Herman W Raadsmal Peter Williamson<sup>1,\*</sup> <sup>1</sup>University of Sydney

Keywords: Comparative Genomics Functional Genomics

A well defined quantitative trait locus (QTL) for important lactation traits (milk fat percentage, protein percentage and milk yield) has been identified on bovine chromosome 6 (BTA6). This QTL was subsequently fine mapped to a 420 kb region on BTA6 containing 6 genes, including ABCG2. Peak location for the QTL was positioned to an interval between the first single nucleotide polymorphism (SNP) of ABCG2 and the second SNP of LAP3.

ABCG2, a member of ATP transporter superfamily, is most recognised for its ability to translocate lipophilic compounds across the plasma membrane, a property which leads to multidrug resistance in chemotherapy. ABCG2 is also highly expressed in stem/progenitor cells while its expression is down-regulated during differentiation. ABCG2, at least in part, is also responsible for the efflux of the specific fluorescent dyes (Hoechst- and Rhodye), thereby identifying the side population (SP) phenotype characteristic of stem/progenitor cells. However, the role of ABCG2 in the initiation and persistency of mammary milk protein expression and in the control of milk composition remain unclear.

To investigate ABCG2 as a candidate gene we used a comparative approach based on RNA expression profiles in mammary tissue. Expression profiles were examined in two mouse models and from public datasets. These were compared to lactation cycle profiles in dairy cattle. The analysis provided clear evidence for differential expression of ABCG2 during mammary gland development. The promoter region of the bovine ABCG2 gene was then targeted to identify potential functional SNPs. Using a pooled DNA sequencing strategy, six SNPs were discovered and validated. A genotyping method was then developed for each SNP using high resolution melt (HRM) analysis. Estimates of population frequency and haplotype association of these SNPs is underway.

WEDNESDAY 15:30 - 15:45 [W4 COMPARATIVE GENOMICS 3]

#### **Use of Comparative Genomics in Identifying Disease Genes in Dogs** Alan N Wilton<sup>1,2,\*</sup> Jeremy Shearman<sup>1</sup>

<sup>1</sup>School of Biotechnology and Biomolecular Sciences, University of NSW

<sup>2</sup>The Clive and Vera Ramaciotti Centre for Gene Function Analysis, University of New South Wales **Keywords:** Comparative Genomics Gene and QTL Mapping

Humans make an excellent model organism for dog disease gene studies. The dog genome sequence can be used to obtain sequence of genes that have been identified in man (or mouse) as good candidates based on similarity in symptoms or characteristics of the disease. Microsatellites can be identified from dog genomic sequence of teh introns and used for homozygosity testing in affected dogs or linkage analysis in pedigrees with affecteds. This will immediately eliminate or identify the candidate as the disease gen in the dog. The causative mutation can then be found be sequencing PCR products designed to contain the exons and splicing sites by using primers designed to intronic sequence (After screening for repeats). We have used this technique to identify mutations for two disorders in Border collie dogs, a nerve degenerative disease and an immune problem, and have begun working on two diseases in Working Kelpies, ataxia and a heart defect. This diseases in the dog can then be used as a model for treatment for the equivalent human disease.

#### TUESDAY 12:30 - 12:45 [T1 COMPARATIVE GENOMICS 1]

**Characterization of divergent immune gene families in a marsupial and a monotreme** Emily Wong<sup>1,\*</sup> Anthony Papenfuss<sup>2</sup> Katherine Belov<sup>1</sup> <sup>1</sup>University of Sydney <sup>2</sup>Walter and Eliza Hall Institute for Medical Research

Keywords: Bioinformatics Comparative Genomics Comparative Genomics

Marsupial and monotreme genomes provide important insight into genome evolution. Using these recently sequenced genomes, we are focusing on studying the evolution of the complex mammalian immune system. Immune genes are known to evolve quickly due to strong selective pressures from pathogens. As a result of this, coding sequences of marsupial and monotreme immune genes tend to be highly divergent from their eutherian orthologs and have proven difficult to identify in the laboratory.

We have used a combination of sensitive and targeted bioinformatics techniques to identify and characterize major immune gene families from the opossum and platypus genomes. Bioinformatic search strategies used to identify these genes include: hidden Markov model searches using HMMer, chaining of HMMer hits, gene prediction using homology information (GenomeScan), use of conserved gene organization (synteny) and phylogenetic analysis. I will discuss the identification of leukocyte receptor complex, Fc receptor and Fc receptor-like genes.

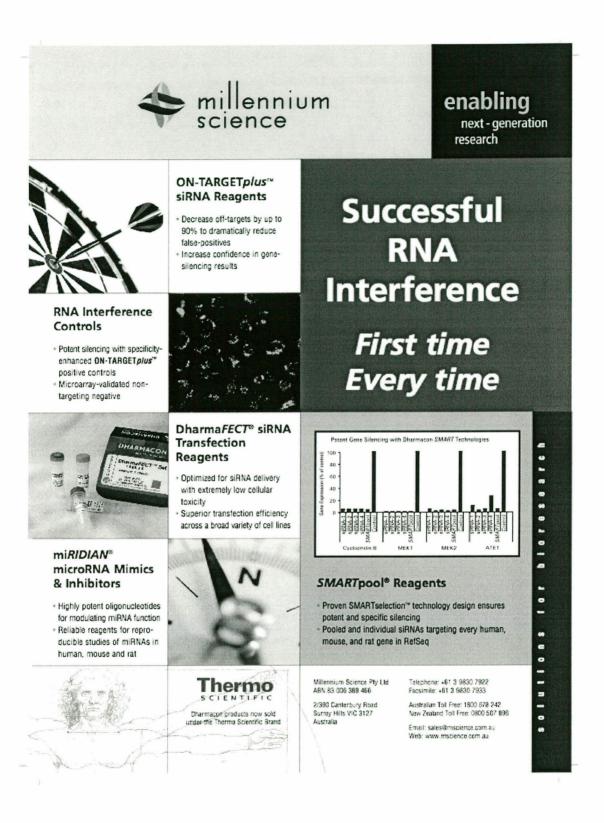
Identification of immune genes from the opossum and platypus has enabled us to map lineage-specific gene expansions and losses during mammalian evolution and deduce the organization of immune genes in ancestral mammals.

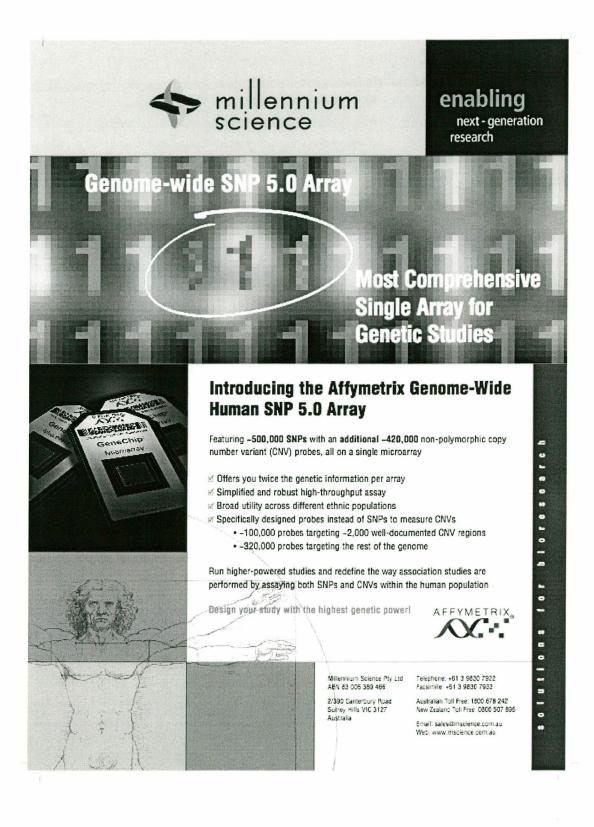
WEDNESDAY 14:00 - 14:15 [W6 FUNCTIONAL GENOMICS 1]

**Deletion of msh-2 in Neurospora: expected phenotype with some surprises** P Jane Yeadon<sup>1,\*</sup> Frederick J Bowring<sup>1</sup> David E A Catcheside<sup>1</sup> <sup>1</sup>School of Biological Sciences, Flinders University **Keywords:** Functional Genomics Comparative Genomics

Msh-2 is a highly conserved protein required for eukaryotic repair of mismatches in DNA. We have deleted the Neurospora msh-2 orthologue in a number of strains. Of 43 octads from an msh-2 deletion homozygote, 6 displayed non-Mendelian segregation at his-3, lys-4 or cot-1. In all cases the marker segregated 5:3, confirming the requirement for Msh-2 in Neurospora mismatch repair. Among 192 random progeny of a mutant cross all 16 genotypes were detected, including those that arose from double or triple crossovers, extremely rare events in a wild type cross. Msh-2 may thus have an unexpected role in crossover interference. Although an msh-2RIP null allele was shown to increase his-3 allelic recombination, we have found this to be true only for his-3 alleles of different wild-type origin, suggesting Msh-2 may regulate recombination between dissimilar sequences. In contrast, preliminary data suggest crossing over in intervals flanking his-3 is unaffected by deletion of msh-2, even when the sequences are known to be substantially divergent.

This work was supported by a grant from the Australian Research Council.







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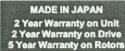


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## Poster Abstracts

THURSDAY 14:00 - 16:00 [PA-F POSTERS]

Genetic structure of blue whales (Balaenoptera musculus) in the Southern Hemisphere Catherine Attard<sup>1,\*</sup> Luciana Möller<sup>1,2</sup> Curt Jenner<sup>3</sup> Peter Gill<sup>4</sup> Naohisa Kanda<sup>5</sup> Micheline Jenner<sup>3</sup> Margie Morrice<sup>4</sup> John Bannister<sup>3</sup> Luciano Beheregaray<sup>1</sup>

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<sup>3</sup>Centre for Whale Research, P.O. Box 1622, Fremantle, WA 6959, Australia

Australocetus Research, c/- Post Office, Narrawong, VIC 3285, Australia

<sup>5</sup>The Institute of Cetacean Research, Tokyo, Japan

Keywords: Conservation Genetics Population Genetics

Whaling has dramatically reduced the abundance of blue whales (Balaenoptera musculus) worldwide. The species is currently classified as endangered by the International Union for the Conservation of Nature and Natural Resources (IUCN). In the Southern Hemisphere two subspecies have been identified; the 'pygmy' blue whale (B. m. brevicauda) in lower latitudes and Antarctic 'true' blue whale (B. m. intermedia) in higher latitudes. The two main Australian feeding aggregations are found in the Perth Canyon, Western Australia and in the Bonney Upwelling, South Australia and Victoria. However, there is limited knowledge about blue whale population structure and subspecies differentiation. In this study we are developing blue whale microsatellite markers, which in combination with mitochondrial DNA (mtDNA) control region sequences will be used to investigate the genetic structure of the two main Australian feeding aggregations. In addition, the novel genetic markers will be used to assess diagnostic genetic differences between the two Southern Hemisphere subspecies. Here we present preliminary results about genetic variation and levels of gene flow in blue whales from the two main Australian feeding aggregations and the Antarctic and discuss the implication of these results on conservation management.

#### THURSDAY 14:00 - 16:00 [PA-F POSTERS]

GEM DB: Griffith Ensembl Mirror Database Kimberly N Begley<sup>1,\*</sup> David C Green<sup>1</sup> Phil Whipps<sup>1</sup> Andrew Lewis<sup>1</sup> <sup>1</sup>Research Computing Services, Griffith University, Queensland, Australia Keywords: Bioinformatics

GEM involves the installation of a local Ensembl database on the Storage Resource Broker (SRB) platform at Griffith University. Ensembl is a joint project between EMBL-EBI and the Sanger Institute consisting of a database containing annotated eukaryotic genomes -- it is a trusted well known bioinformatics resource. Key priority of GEM is to provide secure access to researchers at other institutions to integrate their own research data with the Ensembl dataset. This is achieved using a Distributed Annotation Server (DAS) with Shibboleth -- a middleware product that authenticates users by their home institution user names and passwords. SRB is a client-server middleware that will allow other SRB users to access the Ensembl data without having to maintain their own local copy of it. Users can then query Ensembl data in a high throughput manner using the Perl API to access the data directly from MySQL tables. GEM provides institutions in Australia with a local genome browser that they can query and integrate their own data with securely using the web interface or using SRB.

#### THURSDAY 14:00 - 16:00 [PA-F POSTERS]

### Function and Regulation of the Drosophila melanogaster Cytochrome P450 gene Cyp12d1

Adrian Boey<sup>1,\*</sup> Philip Batterham1 Phillip Daborn<sup>1</sup> Centre for Environmental Stress and Adaptation Research (CESAR), Department of Genetics, Bio21 Molecular and Biotechnology Institute, The University of Melbourne, Victoria, 3010, Australia Keywords: Functional Genomics

Cytochrome P450s are a large family of membrane-bound enzymes with diverse endogenous and xenobiotic-metabolising functions. We are interested in the function and regulation of the Drosophila melanogaster Cyp12d1 gene. Cyp12d1 is arguably one of the most transcriptionally inducible D.melanogaster P450s, responding to a wide range of foreign substances. We have constructed 5'

promoter nested deletion constructs to identify these enhancers and characterise protein binding partners controlling Cyp12d1 expression. Cyp12d1 is expressed in the midgut, Malpighian tubules and fatbody in third instar larvae, known sites of xenobiotic detoxification (WILLOUGHBY et al. 2006). A screen for insecticide resistance showed that Cyp12d1 overexpression using the UAS-GAL4 system confers resistance to the insecticides DDT and Dicyclanil (DABORN et al. 2007), suggesting a xenobiotic metabolising function. A duplication of the Cyp12d1 gene region is present in some populations to give the Cyp12d1-distal copy (Cyp12d1-d') and the Cyp12d1-proximal copy (Cyp12d1-p'); however the significance of this duplication is unknown. We examined Cyp12d1 expression at different D. melanogaster lifestages in a duplicated and non-duplicated strain using quantitative real-time PCR. The highest expression peaks were found during the white prepuape and late pupal stages, indicating some potential endogenous function during pupation. Duplicated lines have approximately twice the amount of Cyp12d1 expression compared to non-duplicated lines, indicating that both gene copies are expressed. We also created transgenic RNA interference (RNAi) fly lines to specifically knock down Cyp12d1 expression (UAS-Cyp12d1 RNAi). No obvious mutant phenotype was seen in flies when UAS-Cyp12d1 RNAi were driven with a GAL4 line driving RNAi expression in a ubiquitous manner and a GAL4 line driving RNAi expression in the midgut, fatbody and Malpighian tubule. This suggests that Cyp12d1 is not essential for development at the levels of knockdown achieved, or that the Cyp12d1 RNAi construct is not being expressed in the correct pattern at the right amounts required to produce a mutant phenotype.

DABORN, P., C. LUMB, A. BOEY, W. WONG, R. FFRENCH-CONSTANT *et al.*, 2007 Evaluating the insecticide resistance potential of eight Drosophila melanogaster cytochrome P450 genes by transgenic over-expression. Insect Biochemistry and Molecular Biology In press. WILLOUGHBY, L., H. CHUNG, C. LUMB, C. ROBIN, P. BATTERHAM *et al.*, 2006 A

WILLOUGHBY, L., H. CHUNG, C. LUMB, C. ROBIN, P. BATTERHAM *et al.*, 2006 A comparison of Drosophila melanogaster detoxification gene induction responses for six insecticides, caffeine and phenobarbital. Insect Biochemistry and Molecular Biology 36: 934-942.

#### THURSDAY 14:00 - 16:00 [PA-F POSTERS]

## A comparative genomics approach to identifying endosymbiont loci associated with early death in insects

Jeremy C Brownlie<sup>1,\*</sup> Manni Sidhu<sup>1</sup> Markus Riegler<sup>1</sup> Scott L O'Neill<sup>1</sup> <sup>1</sup>School of Integrative Biology, The University of Queensland **Keywords:** Pathogens, Parasites & Symbionts Comparative Genomics

Wolbachia are maternally inherited endosymbiont bacteria that commonly infect a range of insects, arachnids and filarial nematodes. Wolbachia strains that infect insects often modify the host's reproduction to increase the number of infected females within a population. A strain of Wolbachia, wMelPop, which naturally infects Drosophila melanogaster is known to severely reduce the lifespan of adult flies. As many mosquito borne diseases, such as dengue and malaria, are transmitted by older mosquitoes changing the age structure of insect vector species has been proposed as a method to reduce or eliminate disease transmission. Understanding how wMelPop reduces the lifespan of D. melanogaster is an important step towards field applications of Wolbachia aimed at modifying mosquito age structures. Cytological inspections of flies infected with wMelpop or a closely related strain that doesn't induce life shortening, suggests that the virulent strain over-replicates within the host's cells, in particular neural tissues. Previous attempts to map the life-shortening trait failed to identify a locus or loci linked to the virulent trait. Using PCR we have successfully amplified, and subsequently sequenced the entire genome of wMelpop. We present our findings from these experiments, describe potential virulence-associated loci and discuss future strategies in light of our findings.

THURSDAY 14:00 - 16:00 [PA-F POSTERS]

**Investigating how a classic reproductive parasite might also act as a mutualist** Jeremy C Brownlie<sup>1,\*</sup> Bodil N Cass<sup>1</sup> Scott L O'Neill<sup>1</sup> <sup>1</sup>School of Integrative Biology, The University of Queensland **Keywords:** Pathogens, Parasites & Symbionts Functional Genomics

Wolbachia are maternally transmitted intracellular bacteria that infect a broad range of invertebrate species. Wolbachia that infect insects modify their host's reproduction to increase the number of infected females within a population, and are considered parasitic. However, a number of Wolbachia strains do not appear to induce any form of reproductive parasitism, yet are able to exist at high frequency in host populations. In the absence of parasitic traits, theory predicts that Wolbachia should confer a fitness advantage to its host. However despite considerable searching no evidence has been

found for any fitness benefit for Wolbachia infected Drosophila. Analyses of recently completed Wolbachia genome sequences revealed that insect Wolbachia are capable of synthesising several metabolic co-factors and vitamins. As such they may be able to provision hosts and positively influence fitness of infected insects. We have tested this hypothesis by rearing D. melanogaster on different nutritionally deficient diets and in some cases found significant positive fitness effects in the presence of the Wolbachia infection. These effects may help explain an unresolved paradox in Wolbachia biology.

#### THURSDAY 14:00 - 16:00 [PA-F POSTERS]

## Understanding the nature of host adaptation for a common endosymbiont through comparative genomics

Jeremy C Brownlie<sup>1,\*</sup> Marcin Adamski<sup>1</sup> Barton Slatko<sup>2</sup> Elizabeth A McGraw<sup>1</sup> <sup>1</sup>School of Integrative Biology, The University of Queensland <sup>2</sup>Molecular Parasitology Division, New England Biolabs, Ipswich, MA 01938 USA Keywords: Pathogens, Parasites & Symbionts Comparative Genomics

Wolbachia pipientis are intracellular bacteria that infect a broad range of arthropod and filarial nematode hosts. Wolbachia strains that infect nematodes are phylogenetically congruent with their host, strictly vertically transmitted, and are required by their hosts for growth and reproduction - typical of many other mutualistic symbioses. In contrast Wolbachia strains that infect insects often form parasitic associations with their hosts. Typically host populations are polymorphic for infection, horizontal transmission of Wolbachia strains occurs between distantly related hosts, and in some instances fitness costs are imposed. These diverse associations form an attractive model for understanding host:symbiont coevolution, yet relatively little is known about the molecular mechanisms that infect the filarial nematode Brugia malayi (mutualist) or Drosophila melanogaster (reproductive parasite) to a closely related outgroup, we identified genes that have experienced diversifying selection in each Wolbachia lineage. An examination of the function of these targeted genes suggests a number of critical molecular mechanisms that underpin the symbiosis and that underlie adaptation to diverse host environments.

THURSDAY 14:00 - 16:00 [PA-F POSTERS] Estimating the propagule size of a recent outbreak Emilie C Cameron<sup>1,\*</sup> Stuart Gilchrist1 John Sved1 <sup>1</sup>University of Sydney Keywords: Population Genetics Evolutionary Genetics

Invasions of nonindigenous pests constitute a growing threat to agriculture and ecosystems due to their potential for severe ecological and economic impacts. The number of individuals in an invading propagule can greatly influence the success of the invasion and has a large impact on the resulting population's level of genetic diversity. Determining the propagule size can aid in the control of an outbreak by indicating the strength of the genetic bottleneck, the mode of introduction and whether the invasion was an isolated event or whether gene flow is continuous. Using a simulation method, seven different statistics were investigated for estimating the propagule size of an outbreak population. For outbreaks originating from populations with high genetic diversity, the number of alleles was a good estimator of propagule size. When, however, the genetic diversity of the source population was already reduced, allele frequency measures, particularly the likelihood of obtaining the outbreak population from the source population, gave more accurate estimates. In neither case was heterozygosity a good estimator of propagule size. Larger sample sizes from the outbreak population were needed to accurately estimate large propagule sizes. Applying this information to a fruit fly outbreak, it was found that just five flies were needed to found the major population in and around Alice Springs. This has led to a severe reduction in the number of alleles, particularly rare alleles in this population. The data strongly suggest that the population originated from a single founder event. As such this population would be ideal to target with the sterile insect technique in a bid to eradicate fruit fly from the area.

#### THURSDAY 14:00 - 16:00 [PA-F POSTERS]

# Water colour and fish evolution: comparative phylogeography of fishes from the major rivers of Amazonia

Georgina M Cooke<sup>1,\*</sup> Luciano B Beheregaray<sup>1</sup> Ning L Chao<sup>2</sup> <sup>1</sup>Macquarie University <sup>2</sup>Universidade do Amazonas

Keywords: Speciation and Phylogeography Evolutionary Genetics

The Amazon is the largest and the most biodiverse river basin on Earth. Encompassing approximately 7 million km2, this complex aquatic landscape harbors large rivers with dramatic differences in water chemistry and colour. According to Endler (1977), isolating geographic barriers are not necessary to promote population diversification. Rather, strong environmental gradients may result in adaptive divergence and speciation, even in the presence of some gene flow. We hypothesize that the different water qualities of the major rivers of the Amazon basin (i.e. whitewater, blackwater and clearwater) act as an environmental gradient that promotes population diversification. To test this, we sampled 56 populations of seven codistributed species of freshwater fish from a vast area that encompasses the four major rivers in Amazonia: Rios Amazonas, Negro, Madeira, and Tapajós. This project uses a combination of molecular markers (mitochondrial DNA (mtDNA), microsatellites and nuclear introns) to achieve high sensitivity in temporal inferences drawn from the genealogical histories. Here, we present and discuss phylogeographic patterns for the seven codistributed species using sequence data from two genes of the mtDNA genome (ATPase 6 and 8). These preliminary results, in conjunction with data from other markers, will provide insight into the relative role of ecological and geographic factors shaping phylogeographic patterns and will be used to elucidate mechanisms that have promoted diversification in Amazonian fishes.

THURSDAY 14:00 - 16:00 [PA-F POSTERS]

### **Evolutionary and Conservation Genetics of Wobbegong Sharks (Orectolobidae)** Shannon Corrigan<sup>1,\*</sup> Luciano B Beheregaray<sup>1</sup>

<sup>1</sup>Macquarie University

Keywords: Evolutionary Genetics Conservation Genetics

Wobbegong sharks (Orectolobiformes: Orectolobidae) are harvested in commercial trap, line and gillnet fisheries on both the East and West coasts of Australia. New South Wales commercial fisheries reported a steady decline in wobbegong landings for the period 1991-2002 and similar trends are emerging for Western Australian fisheries. Although it remains uncertain whether changes in fishing effort have contributed to this trend, these declines have prompted conservation concern for wobbegongs on account of their K-selected life history and ultimate limited capacity to rebound following anthropogenic pressure. In recognition of this concern the IUCN has classified two species of wobbegong as Vulnerable in NSW and Near- Threatened globally. Irrespective of this, research on wobbegong population biology, population structure and dispersal that would contribute to the development of species specific management plans is limited. In addition, taxonomic confusion is widespread within the family Orectolobidae. This study aims to address these issues by conducting a modern, conservation-oriented assessment of the phylogenetic and phylogeographic history, and population genetic structure of wobbegong sharks. Samples obtained from Australia and the Indo Pacific are being used to obtain genetic information from mitochondrial and nuclear DNA markers. These data will be used to reconstruct the evolutionary relationships among wobbegong species, address taxonomic uncertainties, describe patterns of migration at several geographical scales and identify conservation units. Here, we present some of the first results emerging from our study. Namely, genetic evidence for the splitting of a single wobbegong taxon from the Australian East coast into two valid species and a phylogenetic analysis of the family Orectolobidae based on mtDNA and nuclear sequence data. The implications of these results for the conservation and management of these species will be discussed in the context of our ongoing research program.

THURSDAY 14:00 - 16:00 [PA-F POSTERS]

Classification of Expression Profiles based on Visual Graph Topology Analysis David Cho Yau Fung<sup>1,\*</sup> Georgina Lakeland<sup>2</sup> John Galloway<sup>2,3</sup> <sup>1</sup>Independent Investigation, South Strathfield NSW 2136 <sup>2</sup>NetMap Analytics Pty. Limited, St Leonards, NSW 2065 <sup>3</sup>Complex Systems Research Centre, University of Technology Sydney, NSW 2001 Keywords: Bioinformatics Functional Genomics

Relationships in molecular cell biology are naturally mappable to network graphs since they can be constructed from a large collection of binary relationships. Graph topologies emerged from the

integration of biological relationships can often reveal functional modules and genetic regulatory hierarchies. At present, patient classification based on gene expression profiling has mainly relied on statistical methods or quantitative clustering algorithms. The authors proposed that the same objective can be achieved by visually inspecting the topologies of emergent groups. Emergent groups are subgraphs in which the entities are more likely to be connected to each other than those outside the subgraphs. Furthermore, the biological variations between patient subsets can be studied by incrementally analyzing the network topologies formed from the Gene Ontology-to-patient, Gene-to-patient, and Transcription factor-to-Gene relationships. Using microarray data on human chondrogenesis as a case study, the authors intended to demonstrate the feasibility of applying visual graph mining in microarray analysis.

### THURSDAY 14:00 - 16:00 [PA-F POSTERS]

A User Study on Two Gene Ontology Visualization Methods David Cho Yau Fung<sup>1,\*</sup> Kai Xu<sup>2</sup> Seok -Hee Hong1 David Hart<sup>3</sup> <sup>1</sup>School of Information Technologies, The University of Sydney NSW 2006 <sup>2</sup>National ICT Australia Limited, Locked Bag 9013, Alexandria NSW 1435 <sup>3</sup>Axogenic Pty Limited, Australian Technology Park, Eveleigh NSW 1430 Keywords: Bioinformatics Bioinformatics

Introduction: In recent years, Gene Ontology (GO) has been used increasingly as a data mining dimension. Consequently, there have been active research into data mining algorithms for clustering co-expressed genes in the context of GO but not so in GO visualization. Thus, questions such as (1) what are the alternatives to the published methods for visualizing gene cluster-to-GO biological process relationship and (2) what makes a representation effective in assisting the biologist to exercise their analytical reasoning in the context of GO remain unanswered. The research described in this paper represents the authors' intention to fill this analytical gap in microarray data analysis.

Results: The authors contributed two new representations for visualizing the gene cluster-to-GO biological process relationship and an evaluation study that captured the tasks commonly performed by microarray users. The study showed that for most tasks bipartite graph users performed better than their block matrix counterpart. Secondly, it indicated that the design of an effective GO visualization will require in the order of priority (1) the explicit representation of the n-ary cardinality of the gene cluster-to-GO relationship, (2) non-redundant representation of GO terms, and (3) edge crossing minimization. Finally, the study indicated that GO visualization is inadequate for the purpose of investigating biomedical research problems.

Conclusion: This study put into question as to whether reducing visual complexity by using a nongraph modular design can enhance the biologist's analytical reasoning. The evaluation study showed that the biologist's analytical reasoning cannot be enhanced simply by amplifying one's perception on the modularity of gene expression without representing the cardinality of the gene cluster-to-GO relationship faithfully. The design principles suggested above in conjunction with the results from the user evaluation indicated a preference towards bipartite graph rather than block matrix.

#### THURSDAY 14:00 - 16:00 [PG-R POSTERS]

## The cyst-forming nematode, Globodera rostochiensis, has a multipartite mitochondrial genome

Tracey Gibson<sup>1,\*</sup> Vivian Blok<sup>2</sup> Mark Dowton<sup>1</sup> <sup>1</sup>University of Wollongong <sup>2</sup>Scottish Crop Research Institute **Keywords:** Evolutionary Genetics Evolutionary Genetics

We amplified and sequenced one entire mitochondrial subgenome from the cyst-forming nematode, Globodera rostochiensis. Comparison of the noncoding region of this subgenome with those reported previously for Globodera pallida facilitated the design of amplification primers for a range of subgenomes from G. rostochiensis. We then randomly sequenced five subgenomic fragments, each representative of a unique subgenome. This study indicates that the multipartite structure reported for G. pallida is conserved in G. rostochiensis. A comparison of subgenomic organization between these two Globodera species indicates a considerable degree of overlap between them. Indeed, we identify two subgenomes with an organization identical with that reported for G. pallida. However, other of the subgenomes are unique to G. rostochiensis, although some of these have blocks of genes comparable to those in G. pallida. Dot-plot comparisons of pairs of subgenomes from G. rostochiensis indicate that the different subgenomes share fragments with high sequence identity. We interpret this as evidence that recombination is operating in the mitochondria of G. rostochiensis.

#### THURSDAY 14:00 - 16:00 [PG-R POSTERS]

## Differential evolution of classical and non-classical class I genes of the major histocompatibility complex among suids and peccaries

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Keywords: Evolutionary Genetics

The major histocompatibility complex (MHC) is one of the most dynamic regions of the immune system and plays an important role in the development and regulation of immune response in both the adaptive and innate immunity of vertebrates. Despite the domestic pig (Sus scrofa) MHC being well characterized, studies of closely related wild suid (Suidae) and distantly related peccary (Tayassuidae) species have never been attempted. Here we present preliminary data on MHC variation and relationships between suids from Africa, Asia and Europe and peccaries from the Americas. Domestic pig primers were used to target an MHC class Ia gene. Amplicons were cloned and sequenced. DNA and protein databases searches indicated that some of these MHC sequences within suids are similar to the classical MHC class Ia genes while in peccaries they are similar to the non-classical MHC class Ib genes found in Sus scrofa. Phylogenetic analyses are consistent with this, showing two major clades of MHC sequences, comprising of suid class Ia and peccary class Ib. A possible explanation is that the MHC class Ia gene targeted is only present in suids and originated after this lineage and peccaries diverged from the common ancestor. In order to further understand the variation of this immune region and reveal the ancestral sequence of the MHC class Ia/Ib series, more MHC primers will be tested.

#### THURSDAY 14:00 - 16:00 [PG-R POSTERS]

#### Functional characterisation of Cytochrome P450 functions in Drosophila melanogaster by genetic knockout and RNAi

Thomas W. Harrop<sup>1,\*</sup> Tamar Sztall Philip Batterham1 Phillip J. Daborn1 <sup>1</sup>Centre for Environmental Stress and Adaptation Research (CESAR), Department of Genetics, Bio21 Molecular and Biotechnology Institute, The University of Melbourne, Victoria, 3010, Australia Keywords: Functional Genomics

Cytochrome P450s are a large superfamily of genes that are found in all eukaryotic organisms. In insects, the enzymes encoded by P450 genes are involved in many endogenous and exogenous functions such as catabolism and metabolism of signal molecules involved in development, and detoxification of xenobiotics such as insecticides. Despite the obvious importance of P450s in many biological functions, the role of most of the 85 P450s in *Drosophila melanogaster* is not known.

P450 proteins have two redox partners, CPR and Dare, which supply electrons to the P450 during the reaction cycle and are absolutely required to support P450 function. In order to investigate the functions of *D. melanogaster* P450s *in vivo*, RNAi will be used in combination with the *Gal4/UAS* system to knock down the *Cpr* and *dare* genes, and thus eliminate all P450 activity, in specific tissues and life stages.

Following this, a reverse genetics approach will be used to investigate the *in vivo* functions of some *D. melanogaster* P450s. Genes that are identified by the knock down of *Cpr* and *dare* will be studied, along with several P450s that have been chosen by their expression patterns and evolutionary relationships, such as *Cyp49a1* (one of the five mitochondrial P450s in the *D. melanogaster* genome, which is highly conserved over 500 M.Y. of evolution) and *Cyp28a5* (which is ubiquitously expressed in the adult fly, and responds transcriptionally to various stress conditions). The *FLP/FRT* system, in which recombination is induced between target sites that have been inserted in different locations within the genome, will be used to precisely delete regions of the genome containing P450s, without disrupting surrounding genes. Other P450s will be deleted by imprecise *P*-element mobilisation. In cases where neither of these techniques can be applied, standard gene targeting by transgenic RNAi will be used.

#### THURSDAY 14:00 - 16:00 [PG-R POSTERS]

## Ankyrin Domain Proteins: Abundant, Variable And Useful In Understanding The Wolbachia-Insect Symbiosis

Iñaki Iturbe-Ormaetxe<sup>1,\*</sup> Markus Riegler<sup>1</sup> Wolfgang Miller2 Yi San Leong<sup>1</sup> Scott L O'Neill<sup>1</sup> <sup>1</sup>School of Integrative Biology, The University of Queensland, St. Lucia, Brisbane, QLD 4072 <sup>2</sup>Laboratories of Genome Dynamics, Center of Anatomy and Cell Biology, Medical University of Vienna, Währingerstr. 10, A-1090 Vienna, Austria

Keywords: Pathogens, Parasites & Symbionts Functional Genomics

Genes encoding for proteins containing ankyrin (ANK) repeats are particularly abundant in the genomes of the bacteria Wolbachia pipientis, ubiquitous endosymbionts that infect a wide range of arthropods and filarial nematodes. ANK genes are relatively rare in prokaryotes, including related !-proteobacteria, yet the Wolbachia strain that infects Drosophila melanogaster contains >23 such genes (2% of the total number of genes). ANK domains typically mediate protein-protein interactions in other organisms, but their role in Wolbachia is yet unknown. We have previously shown that ANK proteins are extremely variable between Wolbachia strains that induce different reproductive phenotypes in their hosts, such as strains that induce cytoplasmic incompatibility (CI) and those that don't induce CI. CI is a type of embryonic lethality used by Wolbachia to quickly invade insect populations. Despite extensive research into Wolbachia, the molecular basis of CI remains a mystery. The variability of ANK genes between strains that induce different phenotypes is very interesting, as the proteins encoded in these strains are predicted to have different subcellular localizations, interact with different proteins and potentially play different roles in the symbiosis. We also show that ANK genes, in combination with tandem repeats, are extremely useful as polymorphic markers for the typing of closely related Wolbachia strains, and they can be used in evolutionary studies. As part of our ongoing research, we have developed antibodies against the most interesting ANK proteins and we are currently analyzing their localization and gene expression across Drosophila developmental stages and tissues.

THURSDAY 14:00 - 16:00 [PG-R POSTERS] Phylogenetic characterisation of cycad-cyanobacteria symbiosis Marilena Meloni<sup>1,\*</sup> Michelle Gehringer<sup>1</sup> Jasper Pengelly<sup>1</sup> Will Cuddy<sup>1</sup> Paul I Foster<sup>2</sup> Giorgio Binelli<sup>3</sup> Brett Neilan<sup>1</sup> <sup>1</sup>University of New South Wales <sup>2</sup>Queensland Herbarium <sup>3</sup>University of Insubria Keywords: Evolutionary Genetics Conservation Genetics

Cycads represent the most primitive living seed plants enduring for nearly 300 million years. Having recognisable morphological characters intermediate between vascular non-seed plants and flowering seed plants, they constitute a key node in plants evolution. The evolutionary success of cycads can be ascribed to their ability to cope with a variety of environmental threats (survival through drought and fire, resistance to pathogens and predators, etc.). Much of this 'ability' is thought to derive from the symbiotic interaction of these plants with cyanobacteria which colonise superficial "coralloid roots" and fix nitrogen, providing plants with this essential nutrient in exchange for substrates suitable for their heterotrophic growth. Cyanobacterial bioactive secondary metabolites, when taken up by plants, may play a role in pollination strategies and in protective mechanisms against herbivores. This, together with the cyanobionts' capability to respond to host signals and affect the development of the plant, suggest the hypothesis of an ancient association and a possible host-symbiont co-evolution.

The aim of our study is a phylogenetic investigation of the cycad-cyanobacteria symbiosis, to study the extent of co-evolution, by phylogenetic inference, which exists between the cyanobionts and their hosts, as well as the exchange of nutrients and secondary metabolites between plants and their symbiotic cyanobacteria. We isolated several symbiotic cyanobacterial strains from soil and coralloid roots of cycads reference samples (Macrozamia spp.) obtained at the Royal Botanical Gardens in Brisbane, as well as from natural specimens collected in New South Wales and Queensland. We classified cyanobacterial isolates by 16S rDNA and cycads according to chloroplast rbcL gene and several chloroplast intergenic spacers.

An extensive knowledge of association and co-evolution of host-symbiont can clarify the importance of this type of symbiosis for the adaptive potential of plants and will be fundamental to provide genetic and ecological information for management plans of cycads species.

THURSDAY 14:00 - 16:00 [PG-R POSTERS]

Microsatellites in the Yeast Genome

Angelika Merkel<sup>1,\*</sup> Neil John Gemmell<sup>1</sup> <sup>1</sup>University of Canterbury, School of Biological Sciences **Keywords:** Evolutionary Genetics Bioinformatics

Microsatellites (tandemly repeated DNA sequences of 1-6bp) are currently the genetic makers of choice, spanning applications from genetic mapping to population genetics and DNA forensics. Whilst dominantly viewed us neutral markers, their mutational dynamics are of increasing interest as they have been found to play a role in gene expression and adaptive evolution. Various studies have attempted to examine and explain the complex pattern of evolution that occurs at microsatellite loci, but a comprehensive picture is still missing.

In our studies we combine genomic data mining and statistical analysis to explore microsatellite evolution in a genomic context. In particular, we examine the relationship among microsatellites with respect to other genomic elements, i.e. transposons, replication origins, promoter regions etc., in order to investigate their role within the genome. Subsequently, we utilize our findings in phylogenetic comparisons among related yeast species to establish (or not) microsatellite functions in overall genome evolution.

At this meeting we present some preliminary results, showing that there are distinctive high and low density regions of microsatellites and which correlate with known genomic features. We discuss the possible (functional) implications of these associations.

#### THURSDAY 14:00 - 16:00 [PG-R POSTERS]

## Biogeographic Patterns Across Three Tropical Butterfly Genera (Lepidoptera) in the Indo-Pacific

Chris James Muller<sup>1,\*</sup> Luciano B Beheregaray<sup>1</sup> <sup>1</sup>Macquarie University

Keywords: Phylogeny, Biodiversity & Barcoding Speciation and Phylogeography

This work represents the first comparison of evolution and biogeography of several butterfly groups/genera co-occurring in the Indo-Pacific, namely the area comprising all of South East Asia, the Philippines, New Guinea, Solomon Islands and the remainder of the South West Pacific Islands west of Fiji. The taxa used for this study include all species (and most distinctive subspecies) of the genera Cethosia and Charaxes (both Nymphalidae) and all species groups in the genus Delias (Pieridae). The primary aim of the thesis is to reconstruct separate phylogenies for all three groups and determine patterns of radiation and speciation within the region. Sequences from three genes have been used to reconstruct the molecular phylogenies of each genera. Mitochondrial DNA genes sequenced include 650 base pairs (characters) of COI and 970 bp of ND5, while 400 bp of the wingless gene was used to represent nuclear DNA. Preliminary analysis of the sequence data already acquired for Cethosia and Charaxes shows a strong affinity between the reconstructed phylogenetic trees and the interpreted geological evolution of the region. Wallace's Line, separating Borneo and Kalimantan in the north and Bali and Lombok in the south of the Indonesian archipelago, appears to represent a prominent boundary between highly divergent, older taxa in the east occurring in the Australasian region and younger, similar species in the west, in the Oriental region. There is a very strong correlation between the phenotypic and genotypic relationships among species of both Cethosia and Charaxes on either side of this line. Nearly all species of both genera occurring to the east of Wallace's Line are morphologically very distinctive, with long branch lengths between taxa, suggesting long term isolation, while those to the west are of generally similar external facies, with comparatively low genetic diversity between taxa, suggesting very recent speciation.

#### THURSDAY 14:00 - 16:00 [PG-R POSTERS]

**Spawning color divergence reflects local adaptation among sockeye salmon** Kristina M Ramstad<sup>1,\*</sup> Carol A Woody<sup>2</sup> Fred W Allendorf<sup>3</sup> <sup>1</sup>Allan Wilson Centre, Victoria University of Wellington, <sup>2</sup>US Geological Survey, <sup>3</sup>University of

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#### Keywords: Adaptation Population Genetics

Sockeye salmon (Oncorhynchus nerka) display dramatic secondary sexual traits during spawning, including red body color that is carotenoid-based and highly heritable. We examined variation in body and egg color among thirteen sockeye salmon populations in Lake Clark, Alaska and found evidence for adaptive color differences between fish spawning in glacial and non-glacial habitats. Lake Clark

sockeye salmon populations arose from a single founding event between 100 and 400 hundred generations ago and exhibit low genetic divergence at 11 microsatellite loci (FST < 0.024) that is uncorrelated with spawning habitat type. We found a higher frequency of pink females spawning in glacial habitats with high turbidity. The lighter pink rather than red body color is possibly due to reduced sexual selection for red spawning color in highly turbid water with low visibility. Females with lighter pink body color tended to have darker eggs, apparently due to a trade-off in carotenoid allocation between body and egg color in females. In contrast to females, nearly all males exhibited bright red body color in all habitat types. Mean PST (phenotypic divergence among populations) exceeded neutral FST for female body color, but was less than neutral FST for male body color. This suggests (1) that body color differences between fish spawning in glacial and non-glacial habitats could not be explained by genetic drift alone and (2) that selection favors different body colors in glacial (pink) and non-glacial (red) spawning females and red body color among males spawning in both habitat types. Our data provide the first evidence of adaptive color polymorphism among sockeye salmon.

THURSDAY 14:00 - 16:00 [PG-R POSTERS] Love is in the Air: MHC & Mate Choice in the Cunnningham's Skink Vincenzo Repaci<sup>1,\*</sup> Adam Stow<sup>1</sup> Jenny Donald<sup>1</sup> <sup>1</sup>Department of Biological Sciences Macqaurie University Sydney Keywords: Conservation Genetics Evolutionary Genetics

The genetic basis of behavior is rarely studied in lizards but is of considerable importance to both conservation and evolutionary biology. Specifically, the mechanisms of mate choice and inbreeding avoidance behaviors in lizards are largely unknown. However, the role of the major histocompatibility complex (MHC) in mate choice is well documented in mice, men and a range of other species. MHC determined scent has been shown to be important in mate choice with MHC preference facilitating the avoidance of mating betweens close kin in several species.

This study investigates MHC and mate choice for Cunningham's Skink (Egerina Cunninghami), a monogamous species with low dispersal and strong inbreeding avoidance. MHC will be investigated as a possible basis for mate choice within E. cunninghami using a large multigenerational sample with known pedigrees. The analysis of the effect of MHC type on mate choice in an entirely wild population where multigenerational pedigrees have been establish allows for powerful comparisons of MHC genotypes and mating structures.

This study will offer insight into the affects of habitat fragmentation on mate choice and mating systems, increase the knowledge of evolution of MHC/mate choice systems and the plasticity of sexual selective behaviors.

THURSDAY 14:00 - 16:00 [PG-R POSTERS]

## The Unusual Mitochondrial Genome of the Potato-Cyst Nematode, Globodera rostochiensis Angelique Helena Riepsamen<sup>1,\*</sup>

<sup>1</sup>University of Wollongong

Keywords: Evolutionary Genetics Evolutionary Genetics

Animal mitochondrial DNA (mtDNA) genomes generally consist of a single, circular molecule, approximately 16 kilobase pairs (kb) long. Also, they comprise a remarkably conserved and compact content and organisation of 36-37 functional mitochondrial genes, providing valuable genetic markers for determining phylogenetic relationships. However, the mtDNA of a parasitic nematode, Globodera pallida, is multipartite, consisting of at least six minicircle mtDNA molecules (~6.4-9.4 kb), each containing only some mtDNA genes. Further, several minicircles of G. pallida contain overlapping multigenic mtDNA fragments, suggestive of recombination, a process thought absent in animal mtDNA. We completely sequenced a mtDNA molecule of a closely related nematode, Globodera rostochiensis, in two overlapping fragments. Analysis revealed that this was a minicircle ~9.2 kb long, with only seven mitochondrial genes present; four protein-coding genes (cytochrome b, NADH dehydrogenase subunits 3 and 4 and cytochrome oxidase III) and three tRNA genes. Further, only ND3 and COIII code for protein products uninterrupted by frameshift mutations or premature stop codons. However, the mRNA that would be produced by the Cytb and ND4 genes can both conceivably be rescued by a type of RNA editing (insertion of an additional U in a poly-U tract) identified as occurring in another nematode. The gene content and organisation of G. rostochiensis mtDNA contrasts that

observed in typical animal mtDNA. Consequently, the remaining 29-30 mitochondrial genes required for normal metabolic function are likely to be coded for elsewhere, suggesting that the mitochondrial genome of G. rostochiensis may be multipartite, similar to that observed in G. pallida. Further studies are required to determine the extent of multipartism in animal mitochondrial genomes, and in turn the prevalence of recombination in animal mtDNA.

#### THURSDAY 14:00 - 16:00 [PS-Z POSTERS]

**Candidate gene identification by combining QTL mapping with bioinformatics** Pearl Ly<sup>1</sup> Jennifer A Saleeba<sup>1,\*</sup>

<sup>1</sup>School of Biological Sciences, Macleay A12, University of Sydney

Keywords: Bioinformatics Gene and QTL Mapping Pathogens, Parasites & Symbionts Plant Genetics

One of the current challenges in genetics, even in the age of fully sequenced genomes, is that of moving from a linkage map position to the isolation of the gene or genes that encode a trait. In QTL mapping genes encoding products that have an influence on a particular phenotype can be identified to a given confidence interval on a genetic map. While one might expect that it should be simple to move from the QTL confidence interval to the identification of the segment of DNA that encodes the mapped signal in an organism for which the complete genome DNA sequence has been determined, this has often proved to be a frustrating step. We will describe our bioinformatics approach to addressing this issue that uses data about polymorphism, gene expression and published descriptions of gene function.

#### THURSDAY 14:00 - 16:00 [PS-Z POSTERS]

## Phylogeography, conservation genetics and management of estuary perch (Macquaria colonorum)

Kim Shaddick<sup>1,\*</sup> Chris Burridge<sup>2</sup> Dean Jerry<sup>3</sup> Dean Gilligan<sup>4</sup> Luciano Beheregaray<sup>1</sup> <sup>1</sup>Molecular Ecology Lab, Department of Biological Sciences, Macquarie University, Sydney <sup>2</sup>Department of Zoology, University of Otago, Dunedin, New Zealand <sup>3</sup>Marine Biology and Aquaculture, James Cook University, Townsville QLD <sup>4</sup>Narrandera Fisheries Centre, NSW DPI, Narrandera, NSW **Keywords:** Conservation Genetics Population Genetics

Estuary perch (*Macquaria colonorum*) are a poorly studied catadromous fish species distributed in rivers and estuaries along the east coast of Australia. Their range extends from the Richmond River in northern NSW to the mouth of the Murray River in South Australia, and includes the Arthur and Ansons Rivers in northern Tasmania. A catadromous life history is thought to impact on fish population structure, and is usually associated with particular life history attributes such as spawning location and larval dispersal. In this study, we used 396 bp of the mitochondrial DNA (mtDNA) control region to assess population genetic structure in estuary perch along the entire distribution of the species. DNA sequence data from 17 geographically distinct estuary perch populations (n =274) yielded only four mtDNA haplotypes. Very low sequence divergence (0.001) was evident between these haplotypes. The two most abundant haplotypes (A and C) were distributed across the range of the species. However, their frequencies showed markedly distinct latitudinal disjunctions. Haplotype 'A' was more frequent south of the Clyde River, whereas haplotype 'C' was more common in the northern part of the distribution of the species. This pattern resulted in significant population structure (FST = 0.443). The outcomes of this research, combined with our upcoming analysis of estuary perch microsatellite DNA variation, will have implications for fisheries management and for determining the influence of a catadromous life strategy on population structure.

THURSDAY 14:00 - 16:00 [PS-Z POSTERS]

Inferring the Protein Domain Boundary of Discontinuous Domain Using DomainDiscovery and Inter-Residue Contact Interactions

Abdur Rahman Sikder<sup>1,\*</sup> Albert Y Zomaya<sup>1</sup>

<sup>1</sup>University of Sydney

Keywords: Bioinformatics Comparative Genomics

Wetlaufer introduced the classification of domains into continuous and discontinuous. Continuous domains form from a single chain segment and discontinuous domains composed of two or more chain segments. Richardson identified approximately 100 domains in her review. Her assignment was based on the concepts that the domain would be independently stable and/or could undergo rigid-body-like movements with respect to the entire protein. There are now several instances where structurally similar domains occur in different proteins in the absence of noticeable sequence similarity. Possibly

the most notable of such domains is the TIM-barrel. With the increase in the number of known sequences, computer algorithms are required to identify the discontinuous domain of an unknown protein chain in order to determine its structure and function. We have developed a novel algorithm for discontinuous domain boundary prediction based on machine learning algorithm (DomainDiscovery) and inter-residue contact interactions values. We have used 415 proteins, including one hundred discontinuous domain chains for training. There is no method available that is designed solely on sequence based for prediction of discontinuous domain. DomainDiscovery performed significantly well compared to structure based methods like SCOP, CATH and DOMAK.

#### THURSDAY 14:00 - 16:00 [PS-Z POSTERS]

#### Comparative Mapping And Analysis Of Breakpoint Regions Between Ovine And Bovine Chromosomes Using Radiation Hybrid Mapping

Mini Singh<sup>1,\*</sup> Imke Tammen<sup>1</sup> Frank W Nicholas<sup>1</sup> Herman W Raadsma<sup>1</sup>

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Keywords: Comparative Genomics

Comparative maps are used to identify regions of conserved synteny, and to use these regions to populate the map of the less-well-mapped species. Comparison of banded karyotypes of cattle and sheep has shown that the three sheep metacentric chromosomes 1, 2 and 3 are the result of Robertsonian fusions/ fissions between cattle acrocentric chromosomes; 1 and 3, 2 and 8 and 5 and 11, respectively (Evans, 1973; Schnedl and Czaker, 1974; Bunch et al., 1976; Broad and Hill, 1994; Ansari et al., 1993; Ansari et al. 1996; Ansari et al. 1999). A comparative mapping experiment was used to better define the evolutionary breakpoint regions between the syntenic conservation of ovine chromosomes OAR1, 2 and 3 with bovine chromosomes BTA1 and 3, 2 and 8, and 5 and 11, respectively. The experiment included mapping of ovine markers on a bovine radiation hybrid panel (Womack et al., 1997) and inclusion on a bovine integrated map (Nicholas et al., 2004). Seven microsatellite markers (URB006, GRIK1, TEK, BP17, TEXAN20, BMS695 and ILSTS42) were selected to be representative of the evolutionary breakpoint regions of interest. These markers were mapped to OAR1, 2 and 3 but had not been previously mapped on the bovine integrated map. The locations of the seven ovine markers, URB006, GRIK1, TEK, BP17, TEXAN 20, BMS695 and ILSTS42 were confirmed on BTA3 (8417kb), BTA1 (4393kb), BTA8 (33762kb), BTA8 (31413kb), BTA8 (20536kb), BTA5 (1870kb) and BTA5 (0kb), respectively, and new marker orders were defined. The evolutionary breakpoint regions were reduced from regions of approximately 10cM to a region of 3.8cM between URB006 and BM6438 on OAR1, a 5cM region between TEXAN20 and BM81124 on OAR2, and a specific location at 155.7cM for the breakpoint on OAR3.

THURSDAY 14:00 - 16:00 [PS-Z POSTERS] On Extending the Hardy-Weinberg Law Alan Edmund Stark<sup>1,\*</sup> <sup>1</sup>Self-funded

Keywords: Population Genetics Evolutionary Genetics

Hardy's (1908) model showed how genetic variability, that is gene frequencies, can be sustained in a large population. He assumed random union of parents to obtain the familiar squared binomial distribution of autosomal genotypes (Hardy-Weinberg proportions - HWP). It has since been recognised (Li(1988), Stark (2006)) that random mating is a sufficient but not necessary condition for the establishment and maintenance of HWP. Thus HWP have a much more general basis than random mating. Hardy's paper resolved a problem which troubled biologists of the time but he introduced a fallacy relating to random mating which has persisted - whether for reasons of mathematical convenience or lack of awareness random mating is still invoked in much of the genetics literature. This paper gives a simple table, along the lines of Li (1988), which delivers an arbitrary distribution of genotypes from any starting point while maintaining the same gene frequencies. Thus it shows in a more general way than did Hardy how genetic variability can be conserved. Formulae which convert the table of mating frequencies into a canonical form known as Fisher's Identity (FI) are given. This application of FI is useful in that it assigns two sets of values (vectors x and y, say) to parents according to genotype, the same for males and females. Along with x and y is a pair of correlation coefficients (r and s) which give the correlation between mates with respect to x and y. While the use of x and y (and r and s) may appear an unnecessary embellishment of the basic mating table it follows the traditions of quantitative genetics. But it gives a particular insight in that it questions the arbitrary assignment of a single set of additive values used by Li (1988) to explore his model.

#### THURSDAY 14:00 - 16:00 [PS-Z POSTERS]

#### Mating structure and inter-nest relatedness in Polistes wasps Stephen Hoggard<sup>1</sup> Adam Stow<sup>1,\*</sup> Andrew Beattie<sup>1</sup> <sup>1</sup>Department of Biological Sciences, Macquarie University Keywords: Population Genetics Conservation Genetics

Social and eusocial organisms are at increased risk of epidemic outbreaks owing to the typically large colony sizes and generally low genetic variability within colonies. Despite these risks, social and eusociality is common within many different arthropod taxa, and therefore strategies must have evolved to combat these risks. It has recently been shown that antimicrobial compounds and the manipulation of mating systems are associated with sociality and may be employed to reduce the threat of disease. The Polistes genus of wasp exhibit what is considered a 'primitive' form of eusociality, may have more than one mating female and show a large variation in colony size. In this study we use microsatellite markers to examine the mating system and social structure of Polistes wasps. We will then look for associations between colony size and polygamy. We test the hypothesis that multiple mating will be more frequently employed to enrich genetic variation (and potentially lower disease risk) as colony size increases. On broader spatial scales, we will examine dispersal and genetic partitioning to obtain knowledge on the likelihoods of disease transmission and epidemics among colonies. This study will also contribute basic information on the social structure of this common, yet relatively unstudied genus in Australia.

#### THURSDAY 14:00 - 16:00 [PS-Z POSTERS]

Expression Analysis of Cytochrome P450s in Drosophila melanogaster

Henry Chung1 Tamar Sztal<sup>1,\*</sup> Lee Willoughby1 Chris Lumb1 Mohan Sridar1 Philip Batterham 1 Phillip J Daborn<sup>1</sup>

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Keywords: Developmental Genetics Functional Genomics Developmental Genetics Functional Genomics

Cytochrome P450s are enzymes present in most organisms, from single cell prokaryotes to multicellular animals. Insects have on average 100 different cytochrome P450 family members, perform various important biochemical reactions. Although some P450s have been implicated in the detoxification of xenobiotics (toxic plant compounds and insecticides), and others in the synthesis of 20-hydroxecdysone from plant sterols, the function of the majority of insect P450s is unknown. We are examining the expression of all 86 P450s in the sequenced Drosophila melanogaster strain (y; cn bw sp) in 3rd instar larvae (feeding and wandering stages) using RNA in situ hybridization of DIG probes of P450 antisense ORFs . We found that manyP450s are expressed in the midgut, Malpighian tubules and fat bodies, where they might have roles in the metabolism of endogenous and exogenous compounds. We also found that some specific P450s are expressed in specialized tissues such as the corpus allatum and oenocytes, where they could play key roles in controlling biochemical pathways in development.

#### THURSDAY 14:00 - 16:00 [PS-Z POSTERS]

Transposable elements in the tammar wallaby genome

Katherine Thompson<sup>1,\*</sup> Edda Koina<sup>1</sup> Matthew Wakefield<sup>2</sup> Jerilyn Walker<sup>3</sup> Mark Batzer<sup>3</sup> Jennifer Marshall Graves 1 Jerzy Jurka4

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Keywords: Comparative Genomics Bioinformatics

Transposable elements (TEs) are a major feature of mammalian genomes, representing at least 50% of the content in species investigated to date. These elements have often been dismissed as genomic 'junk', but there are now many examples in which TE-derived repetitive DNA plays a substantial biological role. For instance, TEs have taken over roles as promoters, enhancers or silencers and may be involved in DNA replication, localisation and movement. TEs have been well studied in a number of eutherians, but not marsupials, with the first in-depth characterisation having occurred only very

recently in Monodelphis domestica. We undertook a comprehensive analysis of the repetitive element content of 15MB of DNA sequences from the tammar wallaby, (Macropus eugenii), an Australian marsupial. This study identified six lineage specific Short Interspersed Elements (SINEs) termed WALLSIs, as well as twelve non-LTR elements (including some full-length elements with in tact open reading frames), five LTR elements and four DNA transposons. Polymerase chain reaction (PCR) analysis of the lineage specific SINEs was conducted in order to ascertain the phylogenetic distribution of each element in six Australian marsupials, the opossum, a monotreme, and humans. These elements were found to be specific to marsupials, with some limited to Australian marsupials. They also appear to be retrotransposable element (RTE)-dependent or derived. The overall proportions of the major groups of repetitive elements in tammar wallaby were found to be similar to those of the Brazilian short-tailed gray opossum and of humans, with the major exception being the lineage specific SINEs and the level of RTE activity found. The discovery of an ancient marsupial RTE, as well as younger elements, which retain open reading frames, and the group of RTE-derived SINEs indicates that the level of RTE activity in marsupials is much higher than that of eutherians. This study contributes much needed data to the field of mammalian, and specifically marsupial transposable elements.

#### THURSDAY 14:00 - 16:00 [PS-Z POSTERS]

## Mapping the X-linked angiotensin receptor 2 gene and X-Y pairing in cattle, sheep and goats

Graham C Webb<sup>1,\*</sup> Cynthia DK Bottema<sup>1</sup>

<sup>1</sup>Discipline of Agricultural & Animal Science, Roseworthy Campus, University of Adelaide **Keywords:** Comparative Genomics Gene and QTL Mapping

The angiotensin receptor 2 (AGTR2) gene was localized to the terminal band, Xp24, of the short arm of the sub-metacentric X chromosome in cattle, and to the bands Xq33-34, approximately 2/3 along the long arm of the acrocentric X in sheep and goats. Studies of male bovine meiosis, using plain staining and localization of AGTR2 by FISH, showed that it is the long arm of the X that pairs with the Y-chromosome in cattle. By contrast, the human X pairs with the Y at the terminus of the short arm, and this is probably the case in most other placental mammals.

Chromosomal microdissection data of Hassanane et al. 1998 (Chromosome Res. 6, 213-217) indicates that for the cattle X chromosome, there was a unique chromosomal shift with a reinsertion of the centromere into the shifted region. Thus, it is more likely that the cattle X chromosome is derived from the X of sheep/goats rather than vice versa. The combination of all the data indicates that the ovine Y chromosome should pair with the sheep/goat short arm of the X, which, perhaps for this reason, is longer than the short arms of the acrocentric autosomes. We are currently searching for confirmation of this in male meiosis of sheep. The preferred stages of male meiosis for microscopy, late diplotene or diakinesis, are easily found in the mouse but rare in cattle, and we have not yet sighted them in repeated studies of sheep. We are now using younger males and subjecting the cells to various treatments to try to observe late diplotene or diakinesis in sheep.

#### THURSDAY 14:00 - 16:00 [PS-Z POSTERS]

### **Exploring the functional organization of eukaryotic genomes using Gene Ontology** Hugh J French<sup>1</sup> Rohan BH Williams<sup>1,2,\*</sup>

<sup>1</sup>School of Biotechnology and Biomolecular Sciences, The University of New South Wales, <sup>2</sup>The Clive and Vera Ramaciotti Centre for Gene Function Analysis, University of New South Wales **Keywords:** Functional Genomics Bioinformatics

Functionally related gene-clusters are a key feature of prokaryotic genomes. In eukaryotes, recent evidence suggests that gene order is also nonrandom at an intra-chromosomal level, and recent analysis of linkage-disequilibrium (LD) data from the mouse HapMap project has uncovered functional organisation occurring on an inter-chromosomal level. The importance of the latter concept is also highlighted by recent experimental data concerning the extent of intra- and inter-chromosomal association of genomic regions during transcription. Here, we present our development of methods designed to assess the intra- and inter-chromosomal organization of function in genomes, as measured by gene-product annotations in Gene Ontology (GO) Biological Process. Our methods are based on the notion of quantifying the similarity of GO annotations of pairwise combinations of gene-products. These methods provide a conceptually distinct approach to the widely used enrichment/depletion based methods that have recently been used to characterize the local (intra-chromosonal) organization of function using GO. We are exploring the feasibility of representing these results as two-dimensional "maps" of function, that allow identification of functional associations on an intra- and inter-chromosomal level, with statistical support. Once fully developed and characterized, these approaches

will be applicable to any species with appropriate sequence and functional annotation, and could be used on other types of genomic data, to explore the intra- and inter-chromosomal organisation of the functional genome. Supported in part by NHMRC Australia (RBHW).

#### THURSDAY 14:00 - 16:00 [PS-Z POSTERS]

### The influence of male development time on strength of Wolbachia-induced CI expression in Drosophila melanogaster Ryuichi Yamada<sup>1,\*</sup> Kevin D Floate<sup>2</sup> Markus Riegler<sup>1</sup> Scott L O'Neill<sup>1</sup> <sup>1</sup>School of Integrative Biology, The University of Queensland <sup>2</sup>Lethbridge Research Center, Agriculture and Agri-Food Canada Keywords: Pathogens, Parasites & Symbionts Evolutionary Genetics

Wolbachia, maternally inherited bacterial symbionts of invertebrates, are known to infect a broad range of hosts including crustaceans, mites, filarial nematodes, spiders and at least 25% of all insect species. In arthropods, Wolbachia commonly act as reproductive parasites, and manipulate their host's reproduction in a variety of ways including male killing, feminization of genetic males, parthenogenesis induction or more commonly via cytoplasmic incompatibility (CI). It is considered that all these phenotypes provide a reproductive advantage to infected females, thereby allowing Wolbachia to persist and spread into host populations. CI is the most widespread reproductive modification induced in insects by Wolbachia. Expression of CI in Drosophila melanogaster is regarded as quite variable. Published papers typically show that CI expression is weak and often varies between different Drosophila strains and different labs reporting the results. The basis for this variability is not well understood, but is often considered to be due to unspecified host genotype interactions with Wolbachia. We have found that in this species, male development time can greatly influence CI expression. In a given family, males that develop fastest express 100% CI. The "younger brothers" of these males (males that take longer to undergo larval development) quickly lose their ability to express the CI phenotype as a function of development time. No correlation is seen between this effect and Wolbachia densities in testes, suggesting that a more subtle interaction between host and symbiont is responsible. The observed younger brother effect may explain much of the reported variability in CI expression in this species. When male development time is controlled it is possible to obtain consistent high levels of CI expression, which will benefit future studies that wish to use D. melanogaster as a model host to unravel CI mechanisms.

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