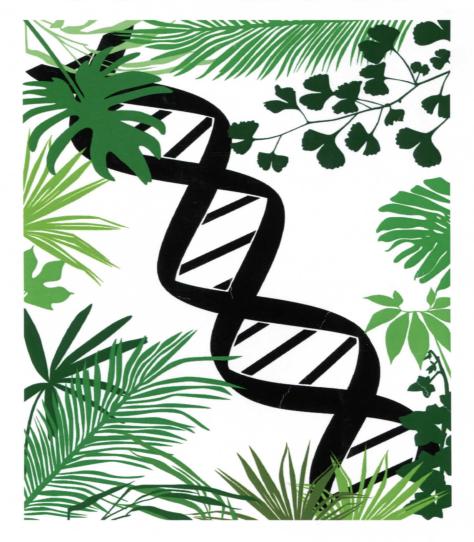
GSA 2009



Brisbane 7-10 July

Programme and abstracts

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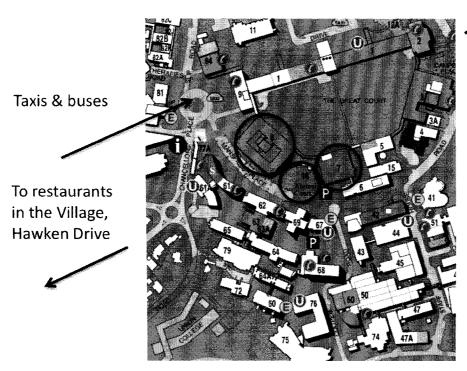
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See the full UQ map provided to you with your registration materials for further information.



UQ CityCat ferry stop

Meeting locations:

Sessions - Buildings 7 & 8.

Registration, plenaries, teas, posters, exhibitors - Building 8

Nearby coffee:

Nano's (very good, but hard to find)- where buildings 60 and 75 meet

Mr. Beans – in between buildings 62 and 63.

Bar Merlo - on the great court in building 2

Computers:

Room 217 in Building 8 (site of posters and rego desk)

iLC rooms, ground floor of building 69, enter where it connects to building 62

Wireless spaces:

Room 217 in Building 8 (site of posters and rego desk)

Science Learning centre inside and tables outside, front of building 67

Nano's coffee (as above)

Great Court

Conference dinner:

Custom's House on the river (399 Queen St, Brisbane City)

or take the CityCat to Riverside stop, walk another few hundred meters down river

GENETICS SOCIETY OF AUSTRALASIA BRISBANE 2009 PROGRAM

7 July 2009

19:00 - 20:30

Opening Mixer

Foyer, UQ Art Museum

8 July 2009

8:00

Registration Opens,

Building 8, room 217

9:00 Welcome Building 8, room 139

ຸ 9:05

Plenary Lecture, Building 8, room 211/39

<u>Prof Greg Gibson</u> Geographical Genomics and the Plasticity of the Human Transcriptome (T1)

10:00

Coffee Break

Building 8, room 217

Session one (chair: D. Ortiz-Barrientos)

> Evolutionary Genetics

Building 7, room 222

- 10:30 <u>David Lambert Mitogenomics of a model penguin (T2)</u>
- 11:00 <u>Steve Chenoweth Single</u> nucleotide contributions to the genetic covariance between a sexually-selected trait and male fitness (T3)
- 11:15 <u>M. Telonis-Scott</u>, R. Hallas , S.W. McKechnie , C.W. Wee , A.A. Hoffmann Selection for cold resistance alters gene transcript levels in *Drosophila melanogaster* (T4)
- 11:30 <u>Stephen W. McKechnie</u>, Mark Blacket, Sue Song, Lea Rako, Xavier P. Carroll, Travis K. Johnson, Louise T. Jensen, Siu F. Lee, Choon W. Wee, Ary A. Hoffmann Climatic adaptation in *Drosophila* involves wing size changes orchestrated by a polymorphic promoter sequence (T5)
- 11:45 Alexandre S. Cristino, Zila L. P. Simoes, Luciano da F. Costa, Denis Anderson, Robyn Russell, John Oakeshott, <u>Charles Claudianos</u> Molecular changes and the evolution of eusocial organisation in the honeybee (T6)
- 12:30 <u>Vidushi S. Patel</u>, Tariq Ezaz, Janine E. Deakin, Jennifer A. Marshall Graves Further evidence to support the new insertional model for globin gene evolution in amniotes: an insight from a reptile (T7)
- 12:45 <u>K.S. Kassahn,</u> V.T. Dang, M.A. Ragan The significance of whole-genome duplications for vertebrate evolution (T8)

12:30

Lunch

Alumni Court

Session Two

<u>Population Genetics</u> (chair: E. McGraw) <u>Building 7, room 222</u>

13:30 <u>Daniel Ortiz-Barrientos</u>
Heterogeneous genomic differentiation during speciation (T9)

(14:00) Alan Wilton, Zi Cong Wu, Sven Warris, Webb Miller, Stephan Schuster Development of SNP markers for examining wild populations (T10)

14:15 Jenny Ovenden, Damien Broderick, David Welch Three unusual mitochondrial genomes from grey mackerel (Scomberomorus semifasciatus) – a case of recombination in animal mitochondrial DNA? (111)

Joel A. Huey, Andrew M. Baker, Jane M. Hughes Evidence for multiple historical colonisations of an endoreic drainage basin by an Australian freshwater fish (T12)

14:45 Thomas R. Buckley, Stick insect (Insecta: Phasmatodea) genetics and environmental change in New Zealand (T13) 15:00 Tanya Llorens, Heidi Nistelberger, Margaret Byrne, David Coates, Colin Yates Fine scale genetic structure and gene flow in Banksia sphaerocarpa var. caesia in the fragmentated agricultural landscape of South-western Australia (T14) 15:15 G.J Houliston, P.B. Heenan, P.J. de

fragmentated agricultural landscape of South-western Australia (T14)
15:15 G.J Houliston, P.B. Heenan, P.J. de Lange, D. Attanyake, A.D. Mitchell Lepidium oleraceum agg. (Brassicaceae) – conservation genetics of Cook's scurvy grass, a critically endangered species complex in New Zealand coastal areas (T15)
15:30 Linda Broadhurst, Andrew Young, David Field, Alec Zwart, Carole Elliott, Gene flow among fragmented Eremophila glabra populations in the western mallee (T16)
15:45 Madeleine Beekman, Ben P. Oldroyd What maintains the natural honeybee hybrid zone in South Africa? (T17)

Session Three

Molecular Genetics (chair: J. Brownlie) Building 7, room 234

13:30 <u>Sureshkumar Balasubramanian</u> Natural variation in *Arabidopsis thaliana*: Flowering and beyond (T18)

14:00 Ivor R. Lee, <u>James A. Fraser</u>
Nitrogen Metabolite Repression: A global regulatory network linking nitrogen scavenging and virulence factors in *Cryptococcus neoformans* (T19)

14:30 Margaret E. Katz, Guancai Yi, Katharyn Sue, Brian F. Cheetham Role of a hexokinase-like protein and a p53-like transcription factor in fungal programmed cell death (T20)

14:45 <u>Richard Burke</u> Genetic regulation of copper homeostasis in *Drosophila* (T21)

Michael B. Thompson Characterisation and uterine expression of vascular endothelial growth factor (VEGF) in reproductive skinks (T22)

15:15) <u>Jeremy R. Shearman</u>, Alan N. Wilton Wapping canine disease genes (T23)

15:30 <u>Matthew J. Wakefield</u>, Alicia Oshlack RNA-seq enables whole genome allele specific expression (T24)

15:45 Andrew H. Lloyd, Jeremy N. Timmis Functional endosymbiotic gene transfer: integration and activation (T25)

THURS

9 July 2009

9:00

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Peter Waterhouse Small RNA mediated processes in plants (T26)

10:00

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Building 8, room 217

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Molecular Evolution (chair: J. Fraser)
Building 7, room 222

10:30 A. Cooper Using ancient DNA to reveal evolutionary processes and the origins of modern phylogeographic patterns (T27)

11:00 <u>Gavin Huttley,</u> Measurement of evolutionary processes using continuous time Markov models (T28)

11:30 R.G. Melvin1, R. Brooks, S. J. Simpson, F. J. Clissold, W. Van Voorhies, M. Lazarou, <u>J.W.O. Ballard</u> Natural History of a two amino acid deletion (T29)

11:45 Mark Dowton, Stephen L. Cameron, Jessica I. Dowavic, Andy D. Austin, Michael F. Whiting⁴ Is the position of mitochondrial tRNA genes selectively neutral? (T30)

12:00 <u>Riepsamen, A.H.,</u> J.R. Barrett, T. Gibson, S.B. Woodworth, M. Dowton Mitochondrial DNA frameshift mutations at homopolymeric sites within COI and COII across the Heteroderidae nematodes (order: Tylenchida) (T31)

12:15 Helen Lindsay, Gavin Huttley Do sex-* specific differences in DNA methylation and transcription levels contribute to male-biased evolution? (T32)

Session 6

Genomics (chair: S. Balasubramanian)
Building 7, room 234

10:30 Camilla M. Whittington, Anthony T. Papenfuss, Wesley C. Warren Katherine Belov Venom gene discovery: beyond the platypus genome. (T33)

11:00 Lee G. Miles, Sally R. Isberg, Travis C. Glenn, Stacey L. Lance, Pauline Dalzell, Peter C. Thomson, Chris Moran A First-Generation Genetic Linkage Map for the Saltwater Crocodile (Crocodylus porosus) (T34)

Thomson, Travis C Glenn, Stacey L. Lance, Chris Moran Quantitative Trait Loci Mapping in Farmed Saltwater Crocodiles (Crocodylus porosus) (T35)

11:30 Veronica J. Murtagh, Paul D. Waters, Yoko Kuroki, Atsushi Toyoda, Asao Fujiyama, Jennifer A. Marshall Graves Sequencing a Y-specific BAC reveals four novel genes on the marsupial Y chromosome (T36)

11:45 Shafagh Al Nadaf, Paul D. Waters, Janine E. Deakin, Edda Koina, Jennifer A. M. Graves X Chromosome inactivation: insights from marsupial mammals (T37)

12:00 Rosie M. Godwin, Jenny Ovenden, Steven Montgomery Telomeres as a potential tool for determining the age of marine invertebrates (T38)

12:15) Hannah S. Bender, Elizabeth P.
Murchison, Hilda A. Pickett, Margaret A. Strong,
Carol W. Greider, Tariq Ezaz, Anne-Maree
Pearse, Roger R Reddel, Jennifer A. Marshall
Graves Telomere length differences between
homologous chromosomes in dasyurid
marsupials – a parental effect? (T39)

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Session 7 Genetic basis of Traits (chair: S. Chenoweth) Building 7 room 222

13:30 Otto Schmidt Inducible tolerance mechanisms in insects (T40)

14:00 Marien de Bruyne, Renee Smart,
Elizabeth Zammit, Jyotika Taneja, <u>Coral G.</u>
Warr A comparative structure-function
analysis to identify ligand-binding regions in
Drosophila Odorant Receptors (T41)

14:30 <u>Benjamin P. Oldroyd</u>, Marcus McHale Expression of genes related to reproduction and pollen foraging in honey bees (*Apis mellifera*) anaesthetised with carbon dioxide (T42)

14:45 Richard D. Newcomb Adventures in mothland: The genetic basis of olfaction in the light brown apple moth, *Epiphyas* postvittana (T43)

(15:00) Vicky Cho, Oscar Luo, Jennifer Henderson, Simon Easteal, Rohan Williams A gene set approach to measuring interindividual variation in gene expression (T44)

15:15) Rachel A. Burt, Marina Carpinelli, Deuglas J. Hilton Investigation of the genetics of hearing loss (T45)

15:30 Alen Faiz, Ben Oldroyd Mapping QTLs for ovary activation in 'anarchistic' honey bees (*Apis mellifera*) (T46)

Session 8
Genomics (chair: E. McGraw)
Building 7, room 234

function of platypus sex chromosomes (T47)
14:00 Janine Deakin, Margaret Delbridge,
Edda Koina, Paul Waters, Amir Mohammadi,
Nerida Harley, Jennifer Graves Mapping
marsupial genomes (T48)
14:15 Michele Dudash, Andrew Young
Genetic rescue in mate-limited populations of
the self-incompatible grassland forb

Butidosis leptorrhynchoides (T49)

74 15 14:30 Paul D. Waters, Natasha Sankovic, Margaret L. Delbridge, Jennifer A. Marshall Graves. Solving mysteries of Y chromosome evolution (T50)

14:45 Erdahl T. Teber, Jason Y. Liu, <u>Sara</u>
Ballouz, Diane Fatkin, Merridee A. Wouters
Comparison of automated candidate gene
prediction systems using genes implicated in
type 2 diabetes by genome-wide association
studies (T51)

14-45 15:00 Denis O'Meally, Tariq Ezaz, Stephen D. Sarre, Arthur Georges, Jennifer A.

Marshall Graves Non-homologous sex chromosomes of birds and snakes share repetitive sequences (F52)

15:15 Asma Al-Huqail, Faisal Al-Saad Identification of Gertain Genes in Four Black Seed (Nigella sativa) Taxa (T53)

/S⋅0015:30 Emily S.W. Wong, Anthony T.

Papenfuss, Marilyn B. Renfree, Richard A.

Gibbs, Katherine Belov Differential

expression in the thoracic and cervical
thymuses of the tammar wallaby (T54)

16:00 16.15 19:00

14.45

15-00

15-15

Coffee Break

Building 8, room 217

Conference Dinner

Friday

10 July 2009

9:00

Plenary Lecture, Building 8, room 139

Andrew G. Clark, Paul Soloway, Xu Wang Biased X-inactivation in the mouse (T55)

10:00

Coffee Break

Building 8, room 217

Session 9

Population Genetics & hair: D. Ortiz-

Barrientos)

Building 7, room 222

10:30 <u>Jack da Silva</u> What do we really know about the molecular population genetics of adaptation? (T56)

11:00 Rachel L. McEvoy, Gavin N. Rees, Shannon K. Dillon, Yvonne M. Parsons¹ Population genetics and adaptation to drought in *Eucalyptus camaldulensis* (T57) 11:15 Melissa A Millar, Margaret Byrne, David Coates Disparate patterns of population genetic diversity, structure and clonality in a critically endangered *Banksia* species (T58)

11:30 Katrina Morris, Julia Beatty, Sonia Cattley, Marilyn Menotti-Raymond, Stephen O'Brien, Katherine Belov Assessing diversity of the MHC in the domestic cat, cheetah and Gir lion using MHC linked microsatellite markers (T59)

11:45 Penelope J. Mills Investigating the presence of a cryptic specices-complex in Apiomorpha minor (Hemiptera: Sternorrhyncha: Coccoidea) (T60)
12:00 Tara Hopley, Andrew Young

Paternity analysis of the autotetraploid invasive tree *Salix cinerea* in south-eastern Australia (T61)

12:15 <u>Jeremy McRae</u>, Yael Salzman, Sara Jaeger, Richard Newcomb Genome-wide analysis of odorant perception (T62)

Session 10

Insect Interactions (chair: C. Claudianos)

Building 7, room 234

10:30 Ross H Crozier, Helge Schlüns, Y Ching Crozier The immune system and insect sociality (T63)

11:00 <u>Jeremy C. Brownlie</u>, Lauren M. Hedges, Scott L. O'Neill, Karyn N. Johnson. Three's a crowd:- *Wolbachia* and pathogen resistance in insects (T64)

11:30 <u>Elizabeth A. McGraw</u>, Luciano Moreira, Andrew Turley and Scott O'Neill A new set of *Wolbachia*-associated host phenotypes in the mosquito, *Aedes aegypti* (T65)

11:45 <u>Eric Caragata</u>, Luciano Moreira, Scott O'Neill, Elizabeth McGraw Improvements to Transcriptional Profilingbased Age grading using in *Wolbachia*infected *Aedes aegypti* (T66)

12:00 <u>Yixin H. Ye</u>, Elizabeth A. McGraw Male-biased survival to infection and immune gene expression in *Drosophila melanogaster* (T67)

12:15 <u>Lauren M. Hedges</u>, Jeremy C. Brownlie, Scott L. O'Neil¹, Karyn N. Johnson Virus protection in *Drosophila melanogaster* by *Wolbachia* (T68)

12:30

Lunch

Alumni Court

13:30 M.J.D. White Address, Building 8, room 139

Jennifer Graves Genomes and sex chromosomes of Australia's weird animals – how my Honours project became my life's work (T69)

14:30 – 15:00 Awards and Conference Close Building 8, room 139

Abstracts (Oral presentations):

T1. Geographical Genomics and the Plasticity of the Human Transcriptome

<u>Greg Gibson</u> University of Queensland

Genome-wide association studies with transcript abundance in peripheral blood samples or derivative cell lines have demonstrated a preponderance of regulatory polymorphisms, also known as eSNPs, which impact the expression of several percent of all genes. Several of these highlight associations that contribute to a variety of disease conditions, but the question arises as to how the associations are affected by the environment. We have addressed the robustness of eSNP associations to plasticity of the transcriptome in the face of different biotic and abiotic challenges faced in different geographic locations. I will describe a gene expression GWAS that controls for population structure and lifestyle, in a comparison of Arab and Amazigh individuals from a city and two villages in southern Morocco. 396 robust cis-eSNP (p<10-08) and 16 transeSNP (p<10-11.5) associations are observed in leukocyte samples obtained from 194 individuals, all of which are consistent across the three sample locations and after controlling for ethnicity and relatedness, despite substantial divergence in the structure of the transcriptome in rural villagers. No evidence for large-effect trans-acting mediators of the pervasive environmental influence is found and instead genetic and environmental factors appear to act in a largely additive manner. I will discuss the implications for the origins of complex disease in human societies undergoing profound transitions.

T2. Mitogenomics of a model penguin

David Lambert
Griffith University

Adélie penguins are remarkable creatures. Perhaps on and off, they have occupied Antarctica for at least hundreds of thousands of years. Their biology is keyed to ice free habitats of a typically frozen continent. I will outline what we know about the late Pleistocene and Holocene distribution of Adélie penguins and present data on the mitogenomic changes in this remarkable species over the last 40,000 years.

T3. Single nucleotide contributions to the genetic covariance between a sexuallyselected trait and male fitness

Steve Chenoweth

School of Biological Sciences, University of Queensland, St Lucia, Australia

Sexual selection is a common and often strong form of selection in natural populations thought responsible for the generation of significant phenotypic diversity. It is therefore surprising that sexually-selected traits have received comparatively little attention in recent efforts to understand the genetic basis of traits underlying fitness differences within natural populations. In this talk, I outline our recent efforts to dissect the genetic basis of sexual display traits in a natural population of the Australian fly, *Drosophila serrata*. This species uses a suite of cuticular hydrocarbons as a pheromonal display system for both mate and species recognition. We are targeting naturally-occurring variation in candidate genes in the insect hydrocarbon biosynthetic pathway (*elongases* and *desaturases*), and examining its association to not only these sexual display traits but also male mating success. I highlight a recent association study performed in a natural population of *D. serrata* using an *indirect* method, useful for traits heavily influenced by environmental variance, where parents are genotyped and groups of offspring are phenotyped. I show how this approach can be used to estimate a SNPs contribution to the genetic covariance between trait and fitness — a parameter fundamental to Robertson's secondary theorem of natural selection.

T4. Selection for cold resistance alters gene transcript levels in *Drosophila* melanogaster.

M. Telonis-Scott ¹, R. Hallas ², S.W. McKechnie ², C.W. Wee ¹, A.A. Hoffmann ¹, ¹Centre for Environmental Stress and Adaptation Research, Department of Genetics, University of Melbourne, Parkville, Vic 3052, Australia. ² Centre for Environmental Stress and Adaptation Research, School of Biological Science, Monash University, Clayton, Vic 3800, Australia

Microarrays have been used to examine changes in gene expression underlying responses to selection for increased stress resistance in *Drosophila melanogaster*, but changes in expression patterns associated with increased resistance to cold stress have not been previously reported. Here we describe such changes in basal expression levels in replicate lines following selection for increased resistance to chill coma stress. We found significant up- or down-regulation of expression in 94 genes on the Affymetrix Genome 2.0 array. Quantitative RT-PCR was used to confirm changes in expression of six genes. Some of the identified genes had previously been associated with stress resistance but no previously identified candidate genes for cold resistance showed altered patterns of expression. Seven differentially expressed genes that form a tight chromosomal cluster and an unlinked gene *AnnX* may be potentially important for cold adaptation in natural populations. Artificial selection for chill coma resistance therefore altered basal patterns of gene expression, but we failed to link these changes to plastic changes in expression under cold stress or to previously identified candidate genes for components of cold resistance.

T5. Climatic adaptation in *Drosophila* involves wing size changes orchestrated by a polymorphic promoter sequence

Stephen W. McKechnie¹, Mark Blacket ¹, Sue Song¹, Lea Rako², Xavier P. Carroll¹, Travis K. Johnson¹, Louise T. Jensen², Siu F. Lee², Choon W. Wee², Ary A. Hoffmann²

¹Centre for Environmental Stress and Adaptation Research (CESAR), School of Biological Sciences, Monash University, Victoria 3800 Australia, ²CESAR, Bio21 Institute, Department of Genetics, The University of Melbourne, Parkville, Victoria, 3010, Australia

Not uncommon among wide-ranging ectothermic species are latitudinal clines in body size – larger individuals being at an advantage in colder climates. While hundreds of genes can now be identified in developmental pathways affecting body size it is of interest to ask which and how many of these are involved in establishing such clines – clines that can form over relatively short periods of evolutionary time. We have focussed on the *D. melanogaster* cold adaptation gene (*Dca*) and used a combination of clinal studies, association studies and targeted gene expression (*Gal4*-UAS) to demonstrate that indel and sequence variation in a response-element-rich region of the promoter affect wing size variation and account for a significant proportion of the latitudinal wing-size cline. These data demonstrate that adaptive shifts can involve single-gene regulatory changes with relatively large effects.

T6. Molecular changes and the evolution of eusocial organisation in the honeybee

Alexandre S. Cristino¹, Zila L. P. Simoes,¹ Luciano da F. Costa,¹ Denis Anderson,² Robyn Russell², John Oakeshott², <u>Charles Claudianos³</u>

¹ Faculdade de Filosofia, Ciencias e Letras, USP, Brazil, ² CSIRO Entomology, Canberra. ³ The Queensland Brain Institute, UQ, Brisbane

The "Honeybee Genome Project" brought new insight how molecular changes encoded in DNA might have contributed to the social evolution of the honeybee. In the current study we calibrate a molecular clock and provide evidence for a common origin of social organization in the social corbiculate group of pollen-collecting bees. Our study shows that the evolution down the Apis mellifera lineage has been associated with an accelerated rate of overall nucleotide substitution and a few quite dramatic amplifications of copy number in particular gene families putatively associated with highly advanced eusocial behaviour. Using this clock we dated divergence times and associated gene duplication events in the bee lineage, including changes to the major royal jelly proteins, olfactory receptors, farnesyl pyrophosphate synthases and certain cytochrome P450s subfamilies. Our molecular data confirm the species is evolving rapidly to a highly specialized ecological niche. Given the diminished gene content of the A. mellifera genome overall, it seems likely that the bursts of gene amplifications we have found contribute to this specialization.

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T7. Further evidence to support the new insertional model for globin gene evolution in amniotes: an insight from a reptile

<u>Vidushi S. Patel¹</u>, Tariq Ezaz², Janine E. Deakin¹, Jennifer A.Marshall Graves¹

¹ARC Centre of Kangaroo Genomics, Research School of Biology, ANU, Canberra ACT 2601, Australia.
²Institute for Applied Ecology, University of Canberra, Bruce ACT 2616, Australia.

The haemoglobin molecule, required for oxygen transportation in the body, is encoded by the members from the α - and β -globin gene clusters. Previously, several complex models were proposed to explain the evolution of these gene clusters. By comparing the genomic context of these genes across vertebrates, we recently proposed a new and much simpler model. We hypothesized that in all vertebrates, there is one ancient region containing the α - and β -globin genes and some flanking genes (MPG-C16orf35-α-β-GBY-LUC7L) that have been conserved for more than 410 MYA. According to our model, a copy of the β-globin gene was inserted into a cluster of olfactory receptors (flanked by RRM1 and CCKBR) in the ancestor of amniotes (>315 MYA), forming the amniote β-globin cluster. However, information on the organization of globin genes in reptiles was missing from our recently proposed model. To fill in this gap, we have now obtained data on globin gene organization in the Australian bearded dragon, Pogona viticeps. We found that in the bearded dragon, the α -globin cluster (α^D - α^A) is flanked by C16orf35 and GBY and is located on a pair of microchromosomes, while the β -globin cluster (β_1 - β_2) is flanked by RRM1 on the 5' end and is located onto the short arm of chromosome 3. However, CCKBR that flanks \beta-globin cluster on the 3' end in other amniotes, is located onto the short arm of chromosome 5, indicating that the separation could have occurred by a translocation between the β and CCKBR genes in the lizard lineage. Therefore, our data from bearded dragon provide further evidence to support our new insertional model on the evolution of β-globin gene cluster

T8. The significance of whole-genome duplications for vertebrate evolution

K.S. Kassahn, V.T. Dang, VT, M.A. Ragan University of Queensland

Vertebrates have experienced several rounds of whole-genome duplication (WGD) early in their evolution. To date, the significance of these events remains controversial, in part because the mechanisms by which WGD contributed to functional evolution or speciation are still incompletely characterised. Here we present an integrated approach to characterise five teleost fish genomes that experienced a WGD after their divergence from tetrapods. Our analyses are based on genome-scale synteny, phylogenetic, temporal and spatial gene expression, and protein sequence data. We find that a minimum of 3-4% of protein-coding loci have been retained in two copies in each of the five fish genomes and many of these duplicates are key developmental genes that function as transcription factors or signalling molecules. Almost all duplicate gene pairs we examined have diverged in spatial and/or temporal expression during embryogenesis. A quarter of duplicate pairs have diverged in function via the acquisition of novel protein domains or via changes in the subcellular localisation of their encoded proteins. We found many examples supporting a model of neofunctionalisation and WGD-duplicates have acquired novel protein domains more often than have single-copy genes. Post-WGD changes at the gene regulatory level were more common than changes at the protein level. We conclude that the most significant consequence of WGD for vertebrate evolution has been to enable more-specialised regulatory control of development via the acquisition of novel spatio-temporal expression domains. We find limited evidence that reciprocal gene loss led to reproductive isolation and speciation in this lineage.

T9. Heterogeneous genomic differentiation during speciation

Daniel Ortiz-Barrientos

The University of Queensland, School of Biological Sciences, St Lucia 4072 Queensland, Australia

Levels of genetic differentiation between populations can be highly variable across the genome. This heterogeneous genomic differentiation may be particularly strong during population divergence and speciation, and may lead to the formation of "islands" of genetic differentiation. Such islands originate because divergent natural selection or reproductive isolation accentuate genetic differentiation in some regions, while the homogenizing effects of gene flow preclude divergence in other regions. Here, we review patterns of genetic differentiation between populations and species and present data from hybrid zone experiments where heterogeneous genomic differentiation is evident despite strong reproductive barriers between species. We discuss this data in light of species concepts, genetic introgression, and the role of ecology and genetics on the origin and persistence of new species.

T10. Development of SNP markers for examining wild populations

Alan Wilton¹, Zi Cong Wu¹, Sven Warris², Webb Miller³, Stephan Schuster³.

¹University of NSW, Australia, ²Hanze University of Applied Sciences, The Netherlands, ³Pennsylvania State University, USA

Study of genetics of wild populations has often relied on microsatellites. The power of SNP data has been shown for human populations. SNPs can be typed cheaply for large numbers of samples. However, the difficulty has been identifying loci that are variable and can be used in SNP assays. Next Generation Sequencing (NGS) allows development of a large panel of SNPs for ~\$50,000. Step 1, use NGS to identify sequences for SNPs. For mammals, a single run on a Solexa or Solid will give 1x coverage of genome. It can be aligned to a reference genome to identify Single nucleotides differences, and indels. If no reference genome, DNA from 2 or more individulas can be combined for sequencing to identify differences or candidate regions for SNPs. Step 2, design capture array for 10 – 50,000 regions containing putative SNPs or candidate regions. Step 3, capture DNA from a number of samples from distribution of your species. Barcode and amplify captured DNA for single run on NGS to identify useful SNPs. Design high throughput SNP genotyping for selected SNPs. We are using this technique in dingoes where we have a reference genome for Step 1 and identified 10,000 high quality SNPs in a 454 run.

T11. Three unusual mitochondrial genomes from grey mackerel (Scomberomorus semifasciatus) - a case of recombination in animal mitochondrial DNA?

Jenny Ovenden¹, Damien Broderick¹, David Welch²

¹Molecular Fisheries Laboratory, Queensland Primary Industries and Fisheries, Brisbane ²Queensland Primary Industries and Fisheries, Fishing & Fisheries Research Centre, JCU, Townsville

Mitochondrial DNA recombination may have recently occurred in a marine fish species, based on sequence from four mitochondrial DNA regions (ATPase, control region, cytochrome oxidase I and cytochrome b) in three grey mackerel individuals (Scomberomorus semifasciatus) from Port Douglas, Queensland. Control region sequence from the three individuals was highly similar to sequence from that gene region obtained from an Australia-wide survey of over 300 other S. semifasciatus individuals. COI sequence in the three Port Douglas fish was identical to that of a closely related species (spotted mackerel, S. munroi). Sequence from the ATPase and cytochrome b regions was dissimilar to 14 (ATPase) and 20 (cyt b) other mackerel and tuna species sequenced in-house or downloaded from GenBank. As recombination is rare in animal mitochondrial genomes, particularly in fish, alternate explanations will be discussed along with possible mechanisms for its occurrence and speculation on the origin of the aberrant ATPase and cytb sequence.

T12. Evidence for multiple historical colonisations of an endoreic drainage basin by an Australian freshwater fish.

<u>Joel A. Huey¹,</u> Andrew M. Baker², Jane M. Hughes¹

¹ Griffith University, Australian Rivers Institute, Griffith School of Environment. Nathan, QLD, Australia. ² Queensland University of Technology, School of Natural Resource Sciences. Brisbane, QLD, Australia. 4001.

The distribution of freshwater species and their genetic structure is partly determined by their ability to colonise and recolonise habitats and to disperse between established populations. In this study we explore the contemporary recolonisation ability of an Australian freshwater fish (Ambassis sp.) in the Lake Eyre basin, central Australia. As this species is found across multiple basins in central and northern Australia, we also aim to explore the mechanisms for historical colonisation of catchments within the basin. We used mtDNA and six newly developed microsatellite loci to elucidate genetic structure both within- and among catchments. We found that Ambassis sp. exhibits weak genetic structure within catchments, suggesting some capacity to recolonise extirpated waterholes after disturbance. Genetic structure revealed that the historical pattern of connectivity among catchments in the Lake Eyre Basin is dramatically different from other species studied in this region, with two divergent clades found in separate catchments. This result, in conjunction with different patterns of genetic structure in each catchment, suggest that Ambassis sp. has colonised Lake Eyre on two separate occasions, from north of the Lake Eyre Basin.

T13. Stick insect (Insecta: Phasmatodea) genetics and environmental change in New Zealand

Thomas R. Buckley

Landcare Research, Private Bag 92170, Auckland, New Zealand

The New Zealand stick insect fauna contains approximately 22 species in 9 genera. These species have radiated into almost all terrestrial habitats from lowland forest to the high alpine zone. Phylogeographic analysis and ecological niche modeling have shown species distributions have shifted dramatically since the Last Glacial Maximum. At least two lowland species were extirpated from the South Island with the exception of possible small refugia in the northeastern South Island. In contrast the alpine species show evidence for long term survival in high altitude and inland regions of the South Island. The alpine species appear to be resilient to extreme environmental conditions. Genomic techniques will be applied to these species to understand the genetic basis of cold tolerance and search for evidence of adaptation and selection.

T14. Fine scale genetic structure and gene flow in *Banksia sphaerocarpa* var. *caesia* in the fragmentated agricultural landscape of South-western Australia.

Tanya Llorens, Heidi Nistelberger, <u>Margaret Byrne</u>, David Coates, Colin Yates Science Division, Department of Environment and Conservation, Perth

Finescale patterns of genetic diversity will be informative regarding evolutionary processes in the historically fragmented landscape system of south-western Australia that has characteristics of high diversity distributed among populations at a regional scale. *Banksia sphaerocarpa* var. caesia is a bird pollinated species that is common throughout the agricultural region of SW WA and occurs on sand in a health woodlands matrix. Finescale genetic diversity and pollen dispersal were investigated in a 20x10 km study site where *B. sphaerocarpa* var caesia occurs in several large reserves and many small vegetation remnants with variable levels of isolation. Genetic diversity using microsatellite markers was moderate to high and not associated with population size, and neighbourhood structure was observed within populations. Significant genetic structure was evident between populations in the north east and south west of the study site despite no current vegetative or geographic isolation barriers between the two groups. A pattern of isolation by distance was also observed in the larger group of populations in the south west suggesting limited gene flow. Assessment of mating system and current gene flow shows high outcrossing and a low to moderate level of pollen immigration into populations in this fragmented landscape.

T15. Lepidium oleraceum agg. (Brassicaceae) – conservation genetics of Cook's scurvy grass, a critically endangered species complex in New Zealand coastal areas.

G.J Houliston¹, P.B. Heenan², P.J. de Lange³, D. Attanyake¹, A.D. Mitchell⁴

¹Ecological Genetics Group, Maanaki Whenua Landcare Research Ltd, ²Allan Herbarium,

Maanaki Whenua Landcare Research Ltd, ³Department of Conservation Terrestrial Research

Unit, Auckland, New Zealand, 4Otago School of Medicine, Christchurch Hospital, Christchurch,

New Zealand

Lepidium oleraceum agg. is a critically endangered complex of coastal Brassicaceae. This has numerous threats to its ongoing survival including herbivory, pathogens and habitat loss through the reduction of seabird colonies. Within this group there is considerable morphological variation and it has been suggested that there are multiple taxa present. A combination of microsatellite and sequence data have been applied to this group to best identify the most critical populations for conservation management, as well as trying to better understand the diversity within the group. Preliminary analysis has identified clear northern, southern and Chatham Island groups which are also supported by morphological evidence. Within these groups there is considerable variation but within populations there is limited variation. This data will be used in the reestablishment of this species on offshore islands in an attempt to best maintain population health.

T16. Gene flow among fragmented Eremophila glabra populations in the western mallee

<u>Linda Broadhurst</u> ¹, Andrew Young ¹, David Field ², Alec Zwart ³ & Carole Elliott ^{1,4} ¹CSIRO Plant Industry, Canberra, Australia. ²Department of Ecology and Evolutionary Biology, University of Toronto, Canada. ³CSIRO Mathematical and Information Sciences, Canberra. 4Fenner School of Environment & Society, ANU, Australia.

Vegetation fragmentation alters both genetic and demographic processes within and among plant populations. We investigated pollen-mediated gene flow among populations of the bird-pollinated plant *Eremophila glabra* in the highly fragmented western mallee of NSW. Adult plants from all populations within a 20 x 20 km grid and progeny from 4 focal sites within the grid were genotyped using SSRs. The hexaploid nature of this species required the development of new software to probabilistically assign paternity to populations within the grid. Our results indicate that: 1) inter-population pollinations are responsible for half of the seed set in the landscape; 2) pollination events commonly occur over scales of up to >5km and; 3) while all populations contribute to the pollen pool, their importance as pollen sources is not equal, with four of the twenty populations surveyed contributing almost 50% of the pollen detected in progeny arrays, while five populations contributed a total of <5%. These results support the view that management of remnant vegetation must explicitly take account of population interactions at the landscape level. Our data also suggest that it is possible to prioritise sites for conservation management based on their relative contribution to landscape level genetic and demographic dynamics.

T17. What maintains the natural honeybee hybrid zone in South Africa?

Madeleine Beekman, Ben P. Oldroyd

Behaviour and Genetics of Social Insects Lab, School of Biological Sciences, University of

Sydney, Sydney.

South Africa is home to two subspecies of honeybee: *Apis mellifera capensis* (the Cape honeybee) and *A. m. scutellata* (the African honeybee) that are separated by a hybrid zone in which both reproduce to produce hybrids. Both subspecies have characteristics that make them highly invasive. The Cape honeybee is unique amongst bees in that its workers are able to produce female offspring without mating (thelytoky). Thelytokous workers can become social parasites of the African bee when moved outside the hybrid zone. Infestation with reproductively active workers ultimately leads to the death of the host colony. The African bee is a proven invader: its introduction into the America's has lead to the extinction of European subspecies. Despite the fact that both honeybee subspecies appear well adapted to outcompete the other, the hybrid zone is stable and neither subspecies is able to increase its range. Previously we suggested that the honeybee hybrid zone is an example of a tension zone in which hybrids are less fit compared with either parental genotype resulting in a barrier to gene flow. Recently we started testing our hypothesised mechanisms that may contribute to the stability of the honeybee hybrid zone. Here we will present our results.

T18. Natural variation in Arabidopsis thaliana: Flowering and beyond.

Sureshkumar Balasubramanian
The University of Queensland, School of Biological Sciences, St. Lucia, QLD 4072.

The mechanistic understanding of complex traits has been challenge for geneticists. Recent advances in statistical approaches towards QTL identification, genome sequence technologies as well as the ease of marker development and large-scale genotyping allow us to identify genes that underlie QTL enabling us look at the mechanisms that give rise to the complex phenotypes. The natural populations of *Arabidopsis thaliana* provide an excellent resource for studying fundamental aspects of complex trait analysis at a molecular level. We have been using wild strains of *Arabidopsis thaliana* to understand the molecular basis of phenotypic variation. I will describe some of our recent findings on the molecular basis of phenotypic variation in life history traits such as flowering time, growth rate and disease resistance.

T19. Nitrogen Metabolite Repression: A global regulatory network linking nitrogen scavenging and virulence factors in *Cryptococcus neoformans*

Ivor R. Lee, <u>James A. Fraser</u> School of Chemistry and Molecular Biosciences, UQ, Brisbane

Nitrogen metabolite repression is a well-studied regulatory phenomenon in fungi belonging to the phylum *Ascomycota*. In the presence of a preferred nitrogen source, transcription of genes required for utilisation of less easily metabolised nitrogen sources is repressed by modulating activity of a family of GATA transcription factors. Here we describe evidence of Nitrogen Metabolite Repression in regulation of uric acid degradation by the pathogenic basidiomycete *C. neoformans*, and reveal that this regulatory paradigm is also a key mechanism controlling coordinated expression of virulence factors such as urease and polysaccharide capsule. Deletion of several predicted GATA-type genes led to identification of *ARE1*, a GATA factor-encoding gene mediating this process. Are1 is essential for utilisation of various nitrogen sources including ammonium—a preferred nitrogen source in fungi—and the predominant nitrogen source in the environmental niche of this pathogen, uric acid. Unexpectedly, Are1 also functions to negatively regulate growth at human body temperature. These studies suggest that in addition to controlling genes for nitrogen acquisition, Are1 is a key regulator of virulence-associated phenotypes.

T20. Role of a hexokinase-like protein and a p53-like transcription factor in fungal programmed cell death

Margaret E. Katz, Guancai Yi, Katharyn Sue, Brian F. Cheetham Molecular and Cellular Biology, University of New England, Armidale, NSW 2351 Australia.

We have identified three genes involved in the response to nutrient depletion in the filamentous fungus, *Aspergillus nidulans*. Two of the genes encode non-catalytic hexokinase-like proteins (HxkC and HxkD) and the third gene product (XprG) belongs to a newly defined class of p53-like transcription factors containing an Ndt80-like DNA-binding domain. Ndt80 is yeast transcriptional activator required for progression through meiosis. Vib-1, a regulator of genes required for programmed cell death in *N. crassa* is a homolog of XprG. Genetic evidence indicates that HxkC and HxkD are negative regulators of XprG. HxkD is a nuclear protein and HxkC is associated with mitochondria. Mitochondrial hexokinases have been shown to block apoptosis in mammals and programmed cell death (PCD) in plants. We have evidence that suggests that HxkC may block XprG-mediated PCD in *A. nidulans*. A hxkC null mutant, but not the hxkC xprG double mutant, exhibits markers of PCD without exposure to any PCD-inducing agent. We have recently found that XprG is localised to the nucleus during growth in nutrient-sufficient medium but most appears to be associated with mitochondria during carbon starvation, consistent with a role in triggering PCD in response to nutrient limitation.

T21. Genetic regulation of copper homeostasis in Drosophila

Richard Burke
Monash University

Copper is an essential micronutrient required as a co-factor for numerous enzymes and animals need to balance their need for copper with its inherent toxicity. We are using the fruit fly *Drosophila melanogaster* to investigate the function of the copper transport genes *Ctr1A*, *Ctr1B* and *DmATP7* and how they regulate copper uptake and efflux in various tissues of the fly including the midgut, eye and developing thorax. These studies involve investigating the expression of these genes, the localization of their gene products and the consequences of loss- and gain-of-function mutations in each gene. We will present data demonstrating how these copper transport genes interact with one another to maintain copper homeostasis and how the localization of each protein is vital for it to function correctly. We will also describe our identification of the vSNARE gene *Syntaxin 5* as a key regulator of copper accumulation. Syntaxin 5 has been implicated in both anterograde and retrograde cellular trafficking and our results provide important evidence of a role for these trafficking pathways in copper homeostasis.

T22. Characterisation and uterine expression of vascular endothelial growth factor (VEGF) in reproductive skinks

Bridget F. Murphy¹, Katherine Belov² and Michael B. Thompson¹

¹ Integrative Physiology Research Group, School of Biological Sciences, The University of Sydney, NSW, Australia. ² Australian Wildlife Genomics Group, Faculty of Veterinary Science, The University of Sydney, NSW, Australia.

While most skinks are oviparous, laying eggs that continue to develop in the nest, some are viviparous (live-bearing), retaining embryos in utero until completely developed. Uterine tissue surrounding embryos becomes increasingly vascular as embryos develop, and vascular proliferation is fastest during the period of prolonged embryo retention in viviparous species. To investigate the molecular mechanisms of uterine angiogenesis, we characterized vascular endothelial growth factor (VEGF) in oviparous and viviparous skinks and used RT-PCR to examine VEGF mRNA expression in the uterus of the viviparous skink, Saiphos equalis. VEGF is one of the most potent of a suite of growth factors up-regulated during the initiation of angiogenesis, and differential expression of VEGF isoforms is associated with uterine angiogenesis in both mammals and birds. Expression of two VEGF transcripts, VEGF₁₉₀ and VEGF₁₆₆, increase in the uterus of *S. equalis* during pregnancy. We also report a very short diffusible VEGF splice variant (VEGF₁₁₁) in *S. equalis*. VEGF₁₁₁ has tissue-specific expression and appears to have multiple roles in reproduction; it is expressed in testis tissue, in uterine tissue during late pregnancy, but is absent in uterus during non-reproductive periods. Such a diffusible VEGF protein may synchronize vascular proliferation in embryonic membranes and surrounding uterine tissue to support an increasing embryonic oxygen demand.

T23. Mapping canine disease genes

Jeremy R. Shearman, Alan N. Wilton

School of Biotechnology and Biomolecular Sciences, University of New South Wales, Sydney, Australia. and Clive and Vera Ramaciotti Centre for Gene Function Analysis, University of New South Wales, Sydney, NSW 2052, Australia

Pure bred dogs exist as genetically isolated highly inbred populations with champion males siring a large proportion of the next generation. This often results in recessive mutations becoming common in a breed. Different breeds often have mutations specific to that breed. This makes dogs a good model for identifying the genetic causes of disease. We have successfully used the candidate gene approach and linkage analysis on microsatellites to identify mutations responsible an immune disorder in the Border collie. The Affymetrix canine SNP array allows whole genome association studies to identify a disease region using a small number of unrelated affecteds and controls. We have used the SNP arrays to map the location of the mutation that causes an ataxia in the Australian Kelpie by identifying a large region of shared homozygosity in affecteds. Using the Nimblegen sequence capture arrays this region was s uenced on the 454 FLX.

T24. RNA-seq enables whole genome allele specific expression

Matthew J. Wakefield, Alicia Oshlack

Bioinformatics, The Walter and Eliza Hall Institute of Medical Research, Parkville.

RNA-seq measures of transcription by fragmentation of mRNA and sequencing on high throughput sequencing platforms. RNA-seq provides an expression measure for each transcript and opportunity for the discovery of novel transcripts, alternative splicing, and measuring allele specific expression at transcribed SNPs. To achieve the promise of RNA-seq an understanding of the data and new statistical methods are critical (Oshlack & Wakefield 2009 Biology Direct 4:14). To validate, optimize, and determine the limits of detection of allele specific expression on the Illumina GAII we have conducted a known truth mixture experiment using tissues from two strains of mice. Our mixture design allows calculation of the true expression level and ratio of alleles for individual samples free from unknown biological effects. We will present results examining the effects of sequence mapping, statistical analysis and sequencing depth on the recovery of the truth using allele specific RNA-seq data. We show that RNA-seq can be used as an accurate and sensitive platform for discovery of allele specific expression with applications in epigenetics, cancer, eQTLs and genomic conflict in hybrids.

T25. Functional endosymbiotic gene transfer: integration and activation.

Andrew H. Lloyd, Jeremy N. Timmis School of Molecular and Biomedical Science, the University of Adelaide, South Australia

DNA transfer from the organelles to the nucleus has been a major driving force of eukaryotic evolution. The vast majority of transferred DNA is non-functional in the nucleus but transferred genes become activated in a small subset of events, resulting in functional gene transfer. We describe experimental observation and molecular characterisation of the steps leading to nuclear activation of a reporter gene that was initially located and active exclusively in the chloroplast genome. Previously a number of lines had been generated in which the chloroplast marker aadA had been transferred to the nucleus where it was not expressed because of its chloroplast-specific (psbA) promoter. We characterised the integrant in one of these lines at the molecular level, determining, for the first time the full sequence of any de novo chloroplast DNA insertion and its flanking nuclear DNA. Independent lines with aadA in 12 different nuclear locations were screened on spectinomycin for functional activation of the gene. From ~1.5 billion cells screened, three plants were regenerated. In one, activation was by acquisition of a nearby nuclear promoter. In a second aadA was expressed by acquisition of a CAAT box and enhancer elements that enabled strong nuclear expression from the chloroplast psbA promoter. Resistance in the third was mitotically unstable and the mechanism of activation was not determined.



T26. Small RNA mediated processes in plants

Peter Waterhouse

School of Biological Sciences, University of Sydney

RNA interference (RNAi) is widely used to silence genes in plants and animals. It is induced by delivery of exogenous dsRNA or self-complementary hairpin (hp) RNA, operates through the degradation of target mRNA by endonuclease complexes, and is guided by ~21nt short interfering (si) RNAs. A similar, endogenously-triggered process regulates the expression of some developmental genes through ~21nt micro (mi) RNAs. In animals, these small RNAs are produced from template RNAs by an enzyme called Dicer, and in plants, such as *Arabidopsis*, there are four different types of Dicer-like (*DCL*) protein, each of which produces small RNAs that direct different functions. We have been examining the roles of these DCLs, and associated proteins, and have developed models for their actions. One intriguing process, that I will be describing, is the ability of hpRNAs to engender signals that can travel long distances within the plant to direct gene silencing in remote tissues.

Plenary

T27. Using ancient DNA to reveal evolutionary processes and the origins of modern phylogeographic patterns

A. Cooper

Australian Centre for Ancient DNA, Earth and Environmental Sciences, University of Adelaide, South Australia 5005.

Ancient DNA studies of Ice Age populations of large vertebrates have revealed a dynamic picture of migrations, extinctions, and localised bottlenecks that comprise the background to phylogeographic patterns in modern populations. Disturbingly, it appears unlikely that accurate evolutionary histories could be inferred using just the distribution and genetic diversity of the modern populations alone. These groups also suggest that there may be significant problems in using the geographic distribution of modern populations to estimate the timing and nature of colonisation events. Ancient DNA data also indicates that taxonomic studies may have considerably under-estimated the extent of morphological plasticity within the fossil records of large mammal species. Multiple morphological variants originally thought to represent species or subspecies level splits have been found to show no genetic isolation at the mitochondrial level. Instead these forms appear to represent temporal and spatial morphs, raising interesting questions about the role of epigenetics in the response of animals to environmental change. To investigate these issues we have performed the first genetic scans for methylcytosine and genome-wide SNP variation in Ice Age vertebrate populations using bison DNA.

T28. Measurement of evolutionary processes using continuous time Markov models

Gavin Huttley

Australian National University

Continuous time Markov process models are among the most popular approaches to understanding the factors affecting genomic diversity. The capacity to flexibly parameterise these models is widely exploited for measuring the relative influence of different processes on sequence divergence. Of particular significance are context dependent substitution models (e.g. dinucleotide or codon models) which allow measuring the influence of sequence neighbourhoods. I will discuss the two main continuous time rate matrix forms used in context-dependent substitution models, their advantages and limitations, and how radically different conclusions may result as a consequence of their different properties. The theory and results I'll present establish that for most use cases, the evolutionary models implemented in popular software packages such as PAML and HYPHY are systematically biased.

T29. Natural History of a two amino acid deletion.

R.G. Melvin1,^{2,3}, R. Brooks ³, S. J. Simpson⁴, F. J. Clissold⁴, W. Van Voorhies⁵, M. Lazarou⁶, J.W.O. Ballard¹,³

¹ School of Biotechnology and Biomolecular Science, University of New South Wales, Sydney 2052, Australia. Rm 207, Swenson Science Building, 1035 Kirby Drive. University of Minnesota – Duluth, Duluth, MN 55812 USA Evolution & Ecology Center, School of Biological, Earth and Environmental Sciences, University of New South Wales, Sydney 2052, Australia. School of Biological Sciences, The Heydon-Laurence Building A08, University of Sydney, NSW 2006 Molecular Biology Program, New Mexico State University, Las Cruces, New Mexico 88003-8001, USA. Department of Biochemistry, La Trobe University, Melbourne 3086, Victoria, Australia

The maintenance of genetic variation under selection remains one of the most persistent and important problems in evolutionary biology. Dobzhansky's 'balance hypothesis' holds that selection can favour the maintenance of genetic polymorphism. There are three established forms of such balancing selection: heterozygote advantage, negative frequency-dependent selection and selection varying in space and time, in which different genotypes have an advantage in different, often fluctuating, environments. Strong lines of genotypic and phenotypic evidence support balancing selection as an important agent for the maintenance of genetic variation. However, aside from sickle-cell anemia and, arguably, abnormal abdomen syndrome in Drosophila mercatorum, there are few examples that mechanistically explain the physiological, biochemical and life-history traits that underpin the maintenance of genetic variation. Here we study a naturally-occurring nuclear genetic polymorphism in a gene known to influence oxidative metabolism in Drosophila simulans. We document far-reaching consequences of this polymorphism in protein structure, protein expression, mitochondrial copy number, biochemistry, metabolism and life history, providing what we believe is the most complete picture yet of the selective forces acting on any locus. Our results also imply that fluctuating selection may maintain this polymorphism.

T30. Is the position of mitochondrial tRNA genes selectively neutral?

Mark Dowton.¹ Stephen L. Cameron^{2,4}, Jessica I. Dowavic¹, Andy D. Austin³, Michael F. Whiting⁴

¹Centre for Biomedical Sciences, School of Biological Sciences, University of Wollongong, NSW, 2522, Australia. ²Australian National Insect Collection and CSIRO Entomology, Black Mountain Laboratories, PO Box 1700, Canberra, A.C.T., 2601, Australia. ³Australian Center for Evolutionary Biology and Biodiversity, School of Earth and Environmental Sciences, University of Adelaide, S.A., 5005, Australia. ⁴Department of Biology, Brigham Young University, Provo, Utah, 84602, USA

We entirely sequenced two hymenopteran mitochondrial genomes, and the major portion of We combined this data with nine previously sequenced hymenopteran three others. This allowed us to infer and analyse the evolution of the 67 mitochondrial genomes. mitochondrial gene rearrangements. All of these involve tRNA genes, while four also involve larger (protein-coding or ribosomal RNA) genes. We find that the vast majority of mitochondrial gene rearrangements are independently derived; a maximum of four of these rearrangements represent shared, derived organizations, while three are convergently derived. The remaining mitochondrial gene rearrangements represent new mitochondrial genome organizations. These data are consistent with the proposal that mitochondrial genome organization is, for the most part, selectively neutral. Nevertheless, some mitochondrial genes appear less mobile than Three mitochondrial tRNAs have not moved (trnG, trnS₂ and trnF) in any of the sequenced genomes. Genes close to the noncoding region are generally more mobile, but only marginally so. An increased rate of mitochondrial gene rearrangement is not tightly associated with the evolution of parasitism. Although parasitic lineages tend to have more mitochondrial gene rearrangements than non-parasitic lineages, there are exceptions.

T31. Mitochondrial DNA frameshift mutations at homopolymeric sites within COI and COII across the Heteroderidae nematodes (order: Tylenchida)

Riepsamen, A.H., J.R. Barrett, T. Gibson, S.B. Woodworth, M. Dowton School of Biological Sciences, University of Wollongong, New South Wales 2522

Mitochondrial genomes (mtDNA) are used extensively in phylogenetics due to their sole maternal inheritance, apparent lack of recombination, and sequence conservation within a taxon. Typically, animal mtDNA have a highly compact gene arrangement with no introns, minimal non-coding sequence, and with intra-individual variation limited to infrequent silent mutations. However, the mtDNA of cyst-forming, plant-parasitic nematodes (Tylenchida: Heteroderidae) is unusual, with recombination apparent in the *Globodera* genus, as well as abundant intra-individual variation. Our research found that representative nematodes across the Heteroderidae have extensive variation in the length of long poly-thymidine tracts within protein-coding genes, such that the reading frame would not be maintained in all gene copies if they were coded for directly. Comparisons with expressed sequence tag (EST) data indicated that this variation was also present at the mRNA level. Further studies are required to determine whether these copies are translated or edited, and what effect they have on the physiology of the cyst nematodes.

T32. Do sex-specific differences in DNA methylation and transcription levels contribute to male-biased evolution?

<u>Helen Lindsay</u>, Gavin Huttley John Curtin School of Medical Research, ANU, Canberra

The phenomenon of male-biased mutation, or more rapid accumulation of mutations in male than in female lineages, results in a relationship between the evolutionary time each chromosome spends in the male lineage and its nucleotide substitution rate. In general, rates of substitution occur according Y-linked > Autosomal linked > X-linked. The male-biased mutation rate is commonly attributed to mutations occurring at replication, as male germ cells undergo continuous replication whereas female germ cells undergo limited replication cycles. However, the incidence of male-biased mutations of clinical importance does not increase linearly with age, suggesting that factors other than replication contribute to male-biased mutation. Using dinucleotide substitution models to account for the influence of local sequence context on nucleotide substitution rates, we have examined differences in the instantaneous substitution rate matrices estimated for autosomal and sex-chromosomal sequence alignments. Our results indicate that substitutions caused by deamination of methylated cytosine are male-biased, and occur at a faster instantaneous rate in transcribed than in intergenic sequences for both chromosome types. Other classes of substitution also exhibited sex-bias, but these effects were much less prominent. These results suggest that methylation and transcription may both play significant and non-independent roles in male-biased evolution.

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T33. Venom gene discovery: beyond the platypus genome.

Camilla M. Whittington¹, Anthony T. Papenfuss², Wesley C. Warren³, Katherine Belov¹

Faculty of Veterinary Science, University of Sydney. ²Bioinformatics Divisions, The Walter and Eliza Institute for Medical Research. ³Genome Centre, Washington University School of Medicine.

The platypus (*Ornithorhynchus anatinus*) is venomous. The exact function of the platypus crural system is unclear, but it appears to be used most commonly as an offensive weapon to assert dominance over other male platypuses during the breeding season, and it can also be used as an effective defensive mechanism against predators. Envenomation can kill dogs, and in humans, produces swelling and immediate and excruciating pain, which cannot be relieved through normal first-aid practices, as well as nausea, gastric pain, cold sweats and lymph node swelling. Even morphine is reported to be mostly ineffective as an analgesic. The unusual symptoms of platypus envenomation suggest that platypus venom may contain clinically useful and potentially novel substances, but to date our knowledge of platypus venom is incomplete. We have used next-generation sequencing of a platypus venom gland cDNA library to aid gene discovery of platypus venom components. A comparison of 454 and Illumina sequencing results will be discussed. Preliminary results for venom gene discovery will be presented. We predict that some of these genes will be useful sources for compounds for the development of novel drugs.

T34. A First-Generation Genetic Linkage Map for the Saltwater Crocodile (*Crocodylus porosus*)

<u>Lee G. Miles</u>¹, Sally R. Isberg^{1,2}, Travis C. Glenn^{3,4}, Stacey L. Lance³, Pauline Dalzell^{1,5}, Peter C. Thomson¹, Chris Moran¹

¹ Faculty of Veterinary Science, University of Sydney, NSW 2006, Australia. ²Porosus Pty Ltd, PO Box 86, Palmerston, NT 0831, Australia. ³Savannah River Ecology Laboratory, University of Georgia, P.O. Drawer E, Aiken, SC 29802, USA. ⁴ Department of Environmental Health Science, University of Georgia, Athens, GA 30602, USA. 5South Eastern Area Laboratory Services, Randwick, NSW 2031, Australia.

Genome elucidation is now in high gear, and whilst genetic maps have been developed for a broad array of species, surprisingly, no such maps exist for a crocodilian, or any other member of the Class Reptilia. Genetic linkage maps are essential tools for the mapping and dissection of complex quantitative trait loci (QTL), and in order to permit systematic genome scans for the identification of genes effecting economically important traits in farmed crocodilians, a comprehensive genetic linage map is necessary. We report the first genetic linkage map for the saltwater crocodile (*Crocodylus porosus*), and indeed for any member of the Class Reptilia. This map identifies fourteen linkage groups, comprising a total of 180 microsatellite loci, with 23 loci remaining unlinked. The estimated female and male recombination map lengths were 1824.1 and 319.0 cM respectively, revealing an uncommonly large disparity in map lengths between sexes (ratio of 5.7:1) confirming previous preliminary evidence of major differences in sexspecific recombination rates in a species that exhibits temperature-dependent sex determination (TSD). This framework map is sufficiently dense to permit systematic whole-genome scans for QTL in farmed saltwater crocodiles, and will be of great utility to future genomic research in the Order Crocodylia.

T35. Quantitative Trait Loci Mapping in Farmed Saltwater Crocodiles (Crocodylus porosus)

Lee G. Miles¹, Sally R. Isberg^{1,2}, Peter C. Thomson¹, Travis C Glenn^{3,4}, Stacey L. Lance³, Chris Moran¹

¹Faculty of Veterinary Science, University of Sydney, NSW 2006, Australia. ²Porosus Pty Ltd, PO Box 86, Palmerston, NT 0831, Australia. ³Savannah River Ecology Laboratory, University of Georgia, P.O. Drawer E, Aiken, SC 29802, USA. ⁴Department of Environmental Health Science, University of Georgia, Athens, GA 30602, USA.

The emerging Australian crocodile industry produces farmed saltwater crocodiles (*Crocodylus porosus*) for the international skin trade. Although genetic improvement remains a relatively novel concept, some producers have begun implementing multi-trait genetic improvement for the selection of superior animals. Unfortunately, traditional selection based on pedigreed phenotypic records are limited by a large generation interval (13 years), coupled with difficult to measure traits. The ability to incorporate gene-marker information into the existing selection strategy through marker assisted selection (MAS) would make early selection decisions possible from day of hatch, increasing the selection intensity, and thereby increasing the rate of genetic gain.

Consequently we have conducted systematic genome scans for three commercially important traits; inventory head length (InvHL), number of scale rows (SR), and juvenile survival, for the existence of Quantitative Trait Loci (QTL) in a commercial population of saltwater crocodiles at Darwin Crocodile Farm, Northern Territory, Australia. This is the first QTL mapping investigation for a crocodilian, or indeed for any member of the Class Reptilia. Mapping of QTL is an important first step towards the identification of genes and causal mutations for commercially important traits, and the development of selection tools for implementation in animal breeding

important traits, and the development of selection tools for implementation in animal breeding programs in the crocodile industry.

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T36. Sequencing a Y-specific BAC reveals four novel genes on the marsupial Y chromosome.

<u>Veronica J. Murtagh</u>¹, Paul D. Waters¹, Yoko Kuroki^{2,3}, Atsushi Toyoda³, Asao Fujiyama^{3,4}, Jennifer A. Marshall Graves¹

¹Comparative Genomics Group, Research School of Biological Sciences, ANU, Canberra. ²RIKEN Advanced Science Institute, Yokohama, Japan. ³National Institute of Genetics, Mishima, Japan. 4National Institute of Informatics, Tokyo, Japan.

The mammalian Y chromosome is a gene poor chromosome with the enormous responsibility of determining maleness, and a functional coherence in male reproduction. The eutherian Y contains only about 45 genes, all but 4 of which derive from an autosomal region added to the X and Y after the divergence of marsupials. The marsupial Y chromosome is hypothesized to represent a minimal mammalian Y, having not fused with this large autosomal region. This minimal mammalian Y provides the opportunity for complete characterization of a mammalian Y, which will provide insight into the original mammal Y and the process of Y degradation. Complete characterization is difficult for the Y chromosomes of humans and other eutherian mammals.

A combination of screening methods were used to obtain a collection of tammar wallaby (*Macropus eugenii*) Y chromosome specific clones from two different male-derived BAC libraries (MEKBa and MEB1). In particular, primers specific to sequence flanking three Y genes (*SRY*, *ATRY* and *RBMY*) were used for PCR-based screening of the MEB1 library, which resulted in isolation of a further five BACs in the gene-containing region of the tammar Y. Here we present the initial sequencing results for one BAC, covering a 174kb region of the tammar Y chromosome containing the putative sex-determining gene *SRY*. Our work has revealed four novel marsupial specific Y genes. Three have partners on the X chromosome (*HCFC1*, *RPL10*, *MECP2*) and a fourth is a partial copy of an autosomal spermatogenesis associated protein (*SPATA2*).

T37. X Chromosome inactivation: insights from marsupial mammals

Shafagh Al Nadaf, Paul D. Waters, Janine E. Deakin, Edda Koina, Jennifer A. M. Graves

The ARC Centre for Kangaroo Genomics, Research School of Biological Sciences, The Australian National University, Canberra ACT 2601, Australia.

Most mammals have an XX female/ XY male sex chromosome system. To balance X gene dosage between females (two Xs) and males (just one X), one X chromosome is transcriptionally silenced in female somatic cells in an epigenetic process called X chromosome inactivation (XCI). In eutherian mammals, such as humans and mice, XCI is proposed to be complete along the X. However, in human there are many genes clustered in regions that escape XCI. Although not as well studied, a less stable and apparently simpler mechanism of XCI is observed in the distantly related marsupial mammals.

In this study we compared aspects of eutherian and marsupial XCI to gain new insights into the mechanisms and evolution of this fascinating phenomenon. Frequency of mono-allelic transcription for 20 BACs, representing 25 X-borne genes spanning the marsupial X, was examined by RNA-FISH in female and male fibroblasts of our model marsupial, the tammar wallaby. With this data we constructed an activity map of the X to reveal if there was a correlation between gene location and XIC status. We also conducted q-PCR on wallaby fibroblast RNA from five males and seven females to determine expression ratios for a subset of our X-linked genes.

Surprisingly, RNA-FISH results indicated that the inactivation status of X-linked genes in marsupial fibroblasts was different between nuclei from the same individual. Furthermore, unlike the human X, which has clusters of XCI escapees, genes that escape XCI on the tammar X appear to be randomly distributed. No significant difference in expression of X-linked genes was observed between male and female fibroblasts, although a trend towards higher expression levels in females was observed. These results might indicate incomplete and locus specific

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T38. Telomeres as a potential tool for determining the age of marine invertebrates.

Rosie M. Godwin¹, Jenny Ovenden¹, Steven Montgomery²

¹Molecular Fisheries Laboratory, Queensland Primary Industries and Fisheries, Brisbane, ²

Cronulla Fisheries Research Centre of Excellence, NSW Department of Primary Industries, PO Box 21 Cronulla NSW.

Knowledge of the age structure of fisheries populations and individual growth rates is crucial to accurate population modelling which is an integral part of fisheries management. Vertebrate fisheries species have hard structures such as otoliths or vertebrae which can be used as evidence for incremental growth and hence age. However, the majority of invertebrate species do not have structures that can be used to infer growth. In humans and many other vertebrate species, decreasing telomere length has been associated with ageing and in the last 5 - 7 years, researchers have been studying telomere length in non - model organisms as a potential molecular tool for age estimation in these species. In this paper I will present preliminary data showing that telomere length and its relationship with age is highly varied amongst different taxonomic groups.

T39. Telomere length differences between homologous chromosomes in dasyurid marsupials – a parental effect?

Hannah S. Bender¹, Elizabeth P. Murchison¹, Hilda A. Pickett², Margaret A. Strong³, Carol W. Greider³, Tariq Ezaz¹, Anne-Maree Pearse⁴, Roger R Reddel², Jennifer A. Marshall Graves¹

¹Research School of Biological Sciences, The Australian National University Canberra, ACT 2601, Australia ²Cancer Research Group, Children's Medical Research Institute, Westmead, NSW 2145, Australia. ³Department of Molecular Biology and Genetics, Johns Hopkins University School of Medicine, Baltimore, MD 21205, USA, ⁴ Department of Primary Industries and Water, Kings Meadows, Tasmania 7249, Australia

Eukaryotic chromosome ends are capped and protected by specialised nucleoprotein structures called telomeres. They play a fundamental role in maintenance of chromosome stability, control of replicative lifespan, and carcinogenesis. Telomere length and maintenance strategies vary between species and cell types, with most continuously proliferating cells employing telomerase to correct for chromosome shortening during cell division. Telomere biology has been well studied in model species such as human and mouse, however relatively little is known about telomere regulation in other mammals. Here we report that the Tasmanian devil (*Sarcophilus harrisii*), an Australian dasyurid marsupial, possesses extremely long telomeres and an unprecedented bimodal distribution of short and long telomeres between homologous chromosomes. A consistent pattern of telomere dimorphism on the X and Y chromosomes in male dasyurids suggests that long telomeres are paternally derived. As yet we do not understand how such long and disparate telomere lengths are regulated, but possible mechanisms of telomere length homeostasis in the Tasmanian devil and closely related marsupials will be discussed.

T40. Inducible tolerance mechanisms in insects

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Otto Schmidt

Insect Molecular Biology, School of Agriculture, Food and Wine, University of Adelaide, Glen Osmond, SA 5064, Australia

In contrast to recessive resistance mechanisms that are based on rare target site mutations, not much is known about inducible metabolic tolerance to synthetic and biopesticides. Since transmission of tolerance to subsequent generations occurs by a maternal effect we have used reciprocal genetic crosses to distinguish multigenic tolerance from recessive resistance mechanisms. Moreover, exposure of insect populations to sub-lethal concentrations of pesticides allowed the measurement of incremental increases in tolerance and the associated fitness costs. We discuss the implications of inducible tolerance and its epigenetic transmission for integrated management and for the design of new resistance management strategies in natural and agricultural ecosystems.

T41. A comparative structure-function analysis to identify ligand-binding regions in *Drosophila* Odorant Receptors

Marien de Bruyne, Renee Smart, Elizabeth Zammit, Jyotika Taneja, <u>Coral G. Warr</u> School of Biological Sciences, Monash University, Australia

In *Drosophila* odours are detected by a large family of odorant receptors (Ors). Individual Ors can respond to a number of odorants, and individual odorants are detected by more than one Or, providing a combinatorial coding system for olfactory information. Little is known about how odorant receptors from any organism bind odorants. We are taking a comparative evolutionary approach to this question. First, we have measured odour responses across eight different *Drosophila* species from eight types of olfactory receptor neurons (ORNs), which usually express single Ors. Interestingly, many response spectra are well conserved across many million years of evolution despite in some cases large changes in Or sequence. Comparison of sequences of orthologous Ors which we have shown remain functionally equivalent has provided detailed information on important functional residues in the Ors. In addition, for several neuron types we have found a significant change in odour response profile between *D.melanogaster* and another species. We are using *in vivo* and *in vitro* assays of Or function to perform structure-function analysis to identify the causative amino acid changes in the Or underlying the ORN response change.

T42. Expression of genes related to reproduction and pollen foraging in honey bees (Apis mellifera) anaesthetised with carbon dioxide

Benjamin P. Oldroyd, Marcus McHale

Behaviour and Genetics of Social Insects Laboratory, School of Biological Sciences, University of Sydney.

Female solitary bees cycle between periods of foraging for pollen and periods of oviposition. In contrast, honeybee workers spend the first part of their life in a 'reproductive' phase in which they feed larvae, followed by a foraging phase. The 'reproductive ground plan hypothesis' (RGPH) posits that this behavioural transition has its origins in an ancient gene network that once regulated the reproductive cycles of solitary insects. Furthermore, the RGPH suggests that these same genes have been co-opted to control the different reproductive phenotypes of queens and workers.

Anesthesia of young honeybee workers with carbon dioxide makes them skip their reproductive phase and begin foraging. Furthermore, workers so treated never forage for pollen. In contrast, treated virgin queens immediately commence egg laying.

We have examined expression of genes related to foraging behaviour and reproductive behaviour in queens and workers. Expression of many of these genes change in response to anesthesia with carbon dioxide in a caste-specific manner. Our findings provide support for a link between foraging behaviour and reproductive behaviour in eusocial insects.

T43) Adventures in mothland: The genetic basis of olfaction in the light brown apple moth, Epiphyas postvittana

Richard D. Newcomb

Plant & Food Research, Auckland, New Zealand. School of Biological Sciences, University of Auckland, Auckland, New Zealand. Allan Wilson Centre for Molecular Ecology and Evolution, New Zealand.

The light brown apple moth (*Epiphyas postvittana*) is a significant horticultural pest in Australia and New Zealand, but also found in Hawaii, the UK and more recently California. We have been developing gene databases and functional genomic tools for the moth, with a long-term goal of developing new sustainable control technologies based on odours and pheromones. From these databases we have found and characterized a number of possible targets on which to base the design of new odour-based control measures, including odorant binding proteins, odorant hydrolases and odorant receptors. Among the odorant receptors we have found two that recognize odours produced by plants, including the important plant signaling compound, methyl salicylate. Using the sequence information from genes involved in olfaction from *E. postvittana* we have been able to isolate orthologues from species of leafroller moths within the New Zealand endemic genera *Ctenopseustis* and *Planotortrix* and to assess their contribution to mate recognition differences and speciation.

T44. A gene set approach to measuring inter-individual variation in gene expression

Vicky Cho¹, Oscar Luo¹, Jennifer Henderson², Simon Easteal², Rohan Williams¹

¹Molecular Systems Biology Group and ²Predictive Medicine Group, Genome Biology Program, John Curtin School of Medical Research, Australian National University, Canberra, ACT

Understanding the origin of differences in global gene expression between individuals, whether arising from either genetic and/or environmental sources, is a problem of great current interest. Regardless of whether genetic and/or environmental factors are identified as influencing gene expression in a given setting, the patterns of gene expression related to inter-individual differences still need to be need to represented and understood, or in other words, how can we best capture or represent how individuals in a study population differ in the patterning of their gene expression changes? Here, we develop the concept of *inter-individual co-variance* to capture this level of variation in gene expression, using expression microarray data from CNS-tissue in 193 human subjects. Rather than attempt to analyse global expression differences between individuals, we focus on analysing inter-individual differences that are present in *gene-sets* (a "gene-set" being a set of genes that share some common property *e.g.* co-regulation, membership of a known signalling pathway, etc). We use principal components analysis to provide direct visualisation of expression variation in gene sets and discuss how this method, in combination with regulatory motif data, can be used to identify *trans*-acting variants using targeted expression QTL (eQTL) approaches.

T45. Investigation of the genetics of hearing loss

Rachel A. Burt, Marina Carpinelli, Douglas J. Hilton

Molecular Medicine Division, Walter and Eliza Hall Institute of Medical Research

Age-related hearing loss (presbycusis) is a significant public health issue and is predicted to become increasingly so given the aging population. Noise, and drug-induced hearing-loss are also common. The molecular mechanisms resulting in these conditions are poorly understood. However, it is clear that apoptosis of cells within the cochlear is often involved. We are in the process of screening a panel of engineered mouse strains for hearing loss using Auditory-evoked Brainstem Response (ABR) testing. Preliminary data indicates that mutations in the intrinsic pathway of apoptosis do in fact lead to hearing loss. In addition, a series of genome-wide ENU mutagenesis screens have been established to search for genes involved in hearing and hearing loss. The first is a screen for hearing loss using an Acoustic Startle Response (ASR) test, and the second is a screen for mice with circling and/or head bobbing behaviour as a sign of vestibular dysfunction. A number of interesting mutations from each of these screens are currently being mapped. An ENU screen for mutations protecting against ototoxic drugs, such as cisplatin and aminoglycoside antibiotics, is also being set up. Through better understanding of the genetics of hearing loss it is hoped that we will identify therapeutic targets for prevention and treatment of presbycusis and environmentally-induced hearing loss.

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T46. Mapping QTLs for ovary activation in 'anarchistic' honey bees (Apis mellifera)

Alen Faiz, Ben Oldroyd

Behaviour and Genetics of Social Insects Laboratory, School of Biological Sciences, University of Sydney

In insect colonies, if a rare worker lays eggs, the laying worker has increased fitness relative to her sterile sisters. Thus the evolution of worker sterility is difficult to explain. To date, no genes directly linked to the regulation of worker sterility have been discovered. Yet such genes must exist, because it is easy to select strains in which workers lay eggs at high frequency. For example, in the 'anarchist' strain of honey bees maintained at Sydney University, about 40% of workers have activated ovaries, compared to less than 1% in wild-type strains.

We have backcrossed an F₁ anarchist x wildtype cross to the anarchistic strain to produce a colony in which workers segregated for the wild-type and anarchist reproductive phenotype. From this population we selected 137 workers showing no signs of ovary activation, and 132 showing extreme ovary activation. These workers have been screened with 100 informative microsatellite markers spread evenly throughout the genome. Markers showing strong statistical association with reproductive phenotype will be discussed.

- SESSION 8

T47. Organisation and function of platypus sex chromosomes

Frank Grützner

School of Molecular and Biomedical Science, The University of Adelaide, 5005, Adelaide, South Australia

Monotreme sex chromosomes have been controversial for decades but recently we have been able to identify the ten sex chromosome system and the completion of the platypus genome project has revealed a large number of genes on X chromosomes and XY shared regions but we still lack information about Y chromosome specific sequences. At first sight the platypus sex chromosome system seems unwieldy and complex but it has already revolutionized our understanding of the origin and evolution of mammalian sex chromosomes. In terms of meiotic organisation, dosage compensation and sex determination the monotreme sex chromosome system is likely to provide similar powerful insights and our current work is focused on understanding these fundamental aspects of sex chromosome biology in monotremes. I will discuss our recent work that shows that the platypus meiotic sex chromosome chains assembles in a remarkably coordinated fashion and present first data on the repeat and gene content of the Y specific regions of platypus Y chromosomes which provides insights into the level of Y chromosomes differentiation and reveals new candidate genes that may have been be part of an ancestral mammalian sex determination and differentiation pathway.

T48. Mapping marsupial genomes

<u>Janine Deakin</u>, Margaret Delbridge, Edda Koina, Paul Waters, Amir Mohammadi, Nerida Harley, Jennifer Graves

ARC Centre of Excellence for Kangaroo Genomics, Research School of Biology, ANU, Canberra

The inclusion of marsupial genome sequences has proven important in comparative genomic studies aimed at answering questions in vertebrate genome evolution. Although genome sequence alone is an extremely valuable resource, the full utility of sequence data is not realised unless the sequence is anchored to chromosomes. The tammar wallaby is the first Australian marsupial to have its genome sequenced. This is a light coverage genome consisting of many small sequence contigs. Assigning each of these contigs to chromosomes would be a laborious and costly undertaking. An alternative approach is to identify large blocks of genes conserved between the opossum and human genomes and to map genes at either end of these blocks in the wallaby. Information from the opossum genome can then be used to infer the location of genes within these blocks, circumventing the need to map every wallaby sequence scaffold. Over 500 genes have now been physically mapped to wallaby chromosomes using this approach. The emergence of Devil Facial Tumour Disease (DFTD) in the Tasmanian devil population has resulted in a third marsupial genome project. By identifying wallaby/opossum conserved gene blocks, we will be able to construct a complete physical map of the devil genome by mapping just 120 genes. With genome organization data for three marsupials, we will be able to trace the events which have shaped the genomes of these species.

T49. Genetic rescue in mate-limited populations of the self-incompatible grassland forb *Rutidosis leptorrhynchoides*

Michele Dudash¹ Andrew Young²

¹ Department of Biology, University of Maryland, College Park, MD 20742, USA, ² CSIRO Plant Industry PO Box 1600 Canberra ACT 2601, AUSTRALIA

Small populations of the grassland forb *Rutidosis leptorrhynchoides* are genetically mate-limited due to low *S* allele numbers at the self-incompatibility locus. This results in low seed set, skewed reproductive fitness distributions and reduced population viability. Glasshouse crossing experiments indicate a substantial mating advantage for inter-population crosses which are more likely to involve novel *S* alleles. Here we report results of a field-based genetic rescue study in which flowers on plants from two small (<200) and two large (>1000) populations were either left to be open-pollinated, were pollinated with pollen from local plants within their respective populations or with pollen from plants from both large and small foreign populations. Results showed that interpopulation crossing can increase seed set in some small populations to levels above that observed from open-pollination. However, results were variable and, when observed, increases in fertilisation success were generally smaller than effects detected in glasshouse experiments. This is probably due to the effect of background pollination in the field situation and variation in source population *S* allele diversity. Nevertheless these data suggest that genetic rescue through introduction of plants with new *S* alleles is a viable option for increasing the reproductive success of small *R. leptorrhynchoides* populations.

T50. Solving mysteries of Y chromosome evolution.

Paul D. Waters, Natasha Sankovic, Margaret L. Delbridge, Jennifer A. Marshall Graves.

Comparative Genomics Group, Research School of Biological Sciences, The Australian National University, CANBERRA, ACT, 0200.

Email: waters@rsbs.anu.edu.au

Most mammals have an XX female/ XY male sex chromosome system, in which the X and Y evolved from an ordinary pair of autosomes via a process of Y degradation after it acquired a testis-determining gene. Loss of genes from the Y resulted in an imbalance of X gene dosage between the sexes, which is compensated for by the transcriptional silencing one X chromosome in the somatic cells of females in a process called X-chromosome inactivation (XCI). Because marsupials have been evolving independently from eutherian mammals for 148 million years, understanding the gene content of a representative marsupial Y chromosome, the expression profile of these Y genes (compared to their X orthologues), and the XCI status of their X orthologues, helps greatly in unravelling mammalian sex chromosome evolution.

Here I describe two novel tammar wallaby Y genes and qPCR of five wallaby X/Y gene pairs. These results revealed that wallaby Y genes are either testis specific, or have ubiquitous expression that is reduced compared to their respective X orthologues, indicating that most wallaby Y genes have either sub-functionalised or are pseudogenizing. I also examined the XCI inactivation status of the X orthologues of these Y genes; this showed that marsupial XCI is rather leaky and that these genes could be in transition from non-dosage compensated to dosage compensated. Interestingly, of the nine genes retained on Y chromosomes from the proto-Y, all but one of their X-linked partners have an ancestral role in the vertebrate brain or CNS. This is in contrast to genes from an autosomal region that was added to the sex chromosomes of the eutherian ancestor; of the 15 genes retained on eutheran Y chromosomes from this added region, only one is implicated in the vertebrate CNS.

T51. Comparison of automated candidate gene prediction systems using gene implicated in type 2 diabetes by genome-wide association studies

Erdahl T. Teber¹, Jason Y. Liu¹, <u>Sara Ballouz¹</u>, Diane Fatkin^{1,2}, Merridee A. Wouters^{1,2}

¹ The Victor Chang Cardiac Research Institute, Darlinghurst ² School of Medical Sciences, UNSW, Sydney

Automated candidate gene prediction systems allow geneticists to hone in on disease genes more rapidly by identifying the most probable candidate genes linked to the disease phenotypes under investigation. Here we assessed the ability of eight different candidate gene prediction systems to predict disease genes in intervals previously associated with type 2 diabetes by benchmarking their performance against genes implicated by recent genome-wide association studies. Using a search space of 9556 genes, all but one of the systems pruned the genome in favour of genes associated with moderate to highly significant SNPs. Even when confronted with challengingly large intervals, the candidate gene prediction systems can successfully select likely disease genes and filter statistically less-well-supported genetic data. We suggest consensus approaches fail because they penalize novel predictions made from independent underlying databases. To realize their full potential further work needs to be done on prioritization and annotation of genes.

T52. Non-homologous sex chromosomes of birds and snakes share repetitive sequences

<u>Denis O'Meally</u>¹, Tariq Ezaz^{1,2}, Stephen D. Sarre², Arthur Georges¹ and Jennifer A. Marshall Graves¹

¹ Research School of Biology, Australian National University, Canberra, ² Institute for Applied Ecology, University of Canberra

Snake sex chromosomes provided Susumo Ohno with the material on which he based his theory of how sex chromosomes differentiate from autosomal pairs. Like birds, snakes have a ZZ male:ZW female sex chromosome system, in which the snake Z is a macrochromosome much the same size as the bird Z. However, the gene content shows clearly that the snake and bird Z chromosomes are completely non-homologous. We are investigating the distribution of repetitive DNA on the sex chromosomes of a variety of Australian snakes using FISH. We have found that introns of genes that are autosomal in snakes but Z-borne in chicken show varying degrees of amplification on the degenerated W of derived snakes. Interestingly, a similar pattern is observed when a chicken W chromosome paint is hybridised to snake metaphases. This implies a closer relationship between bird and snake sex chromosomes than their putative independent origins 220-285MYA. Loss of genes on the degenerating W also leads to a dosage imbalance of sex-linked genes. We are investigating dosage compensation in snakes using quantitative RT-PCR.

T53. Identification of Certain Genes in Four Black Seed (Nigella sativa)Taxa.

Asma Al-Hugail, Faisal Al-Saad Botany and Microbiology Dept., College of Science, King Saud University

A comprehensive study was conducted to compare DNA fingerprints of four Taxa of black seeds Nigella sativa L. The results have shown that there are several genetic differences between these different black seeds Taxa, which could be considered as genotypic characteristics and lead to classifying them as varieties under the sativa species. To study the DNA fingerprinting of these Taxa, the ISSR method was employed in the PCR technique to determine the levels of polymorphism between their genetic makeups. The obtained banding pattern indicated a high level of polymorphism. When the percentages of dissimilarity between them were computed, it was revealed that they ranged between 21.5-36.3 %. Such a relatively high level of polymorphism substantiated the objectives of the present study which supposed that black seeds grown in the different localities in the world over time have undergone genetic changes to the level that could make them different varieties. In addition to the DNA fingerprinting a comparison between some genes of the four black seeds Taxa was conducted. Twenty four genes representing 24 different enzymes were selected and scanned via PCR technique using suitable SSR primers. The obtained results, thus, have shown some changes in the genetic structure of some of these genes.

T54. Differential expression in the thoracic and cervical thymuses of the tammar wallaby

Emily S.W. Wong^{1,2}, Anthony T. Papenfuss^{2,3}, Marilyn B. Renfree^{2,4}, Richard A. Gibbs⁵ and Katherine Belov^{1,2}

¹Faculty of Veterinary Science, University of Sydney, NSW 2006, Australia ²ARC Centre of Excellence in Kangaroo Genomics ³Bioinformatics Division, The Walter and Eliza Hall Institute for Medical Research, Parkville, Victoria 3052, Australia ⁴Department of Zoology, The University of Melbourne, Victoria 3010, Australia ⁵Human Genome Sequencing Center, Department of Molecular and Human Genetics, Baylor College of Medicine, One Baylor Plaza, Houston, TX, 77030, USA

The thymus is a primary lymphoid organ with a critical role in the development of the immune system and development and maturation of T cells. Diprotodont marsupials are unusual because they have two thymuses: a pair of thoracic thymuses (found in all mammals) and a larger pair of cervical thymuses. To understand the functional differences between these organs we used RNAseq to explore the transcriptomes of both pairs thymuses of a 120 day old tammar wallaby. We confirmed that both thymuses have gene expression profiles that are consistent with T-cell development. However, 58 transcripts were significantly differentially expressed between the thymuses (p<0.05). The most highly differentially expressed genes include tissue-specific antigens, non-coding RNAs, transcription factors as well as genes involved in epigenetic regulation. Interestingly, muscle- and heart-related genes were highly expressed by the cervical thymus but not by the thoracic. This study has confirmed that both sets of thymuses in marsupials appear to be functional and further analysis will provide clues into the complex regulation of the thymic environment in mammals.

T55. Biased X-inactivation in the mouse

Plenary Andrew G. Clark¹, Paul Soloway², Xu Wang¹

¹ Dept. of Molecular Biology & Genetics, Cornell University, Ithaca, NY. ² Division of Nutritional Sciences, Cornell University, Ithaca, NY

X-inactivation in female eutherian mammals has long been considered to occur at random with respect to the parent of origin of the two X chromosomes. Methods for scoring allele-specific differential expression with a high degree of accuracy have recently motivated a quantitative reassessment of the randomness of X inactivation. RNA-seq applied to the transcriptome of reciprocal F1 mice provided a clear X-chromosome-wide bias in expression, consistent with a degree of non-randomness of X-inactivation. We applied Pyrosequencing to multiple gene targets of brain cDNA samples from multiple reciprocal F1 mice, and discovered a small but consistent and highly statistically significant excess tendency to under-express the paternal X chromosome. This apparent bias toward paternal X inactivation is reminiscent of marsupials, suggesting that there may be retained an evolutionarily conserved imprint driving the bias. At present it is not clear whether the bias is driven by incomplete erasure of the paternal X imprint (e.g. meiotic sex chromosome inactivation), or whether the signal is totally erased and there follows a bias in the X-inactivation process itself. We also present a method to identify and confirm genes that escape X inactivation in normal mice by directly comparing the allele-specific expression ratio of multiple X-linked genes in multiple individuals. Evolutionary consequences of the biased X-inactivation will be explored.

T56. What do we really know about the molecular population genetics of adaptation?

Jack da Silva

School of Molecylar and Biomedical Science, University of Adelaide, Adelaide

For most of the history of population genetics, the development of theory outpaced the generation of data. Now, with abundant molecular sequences, we should have ample data to test theory. And yet, in the case of the molecular basis of adaptation, we still have very few empirical results for even the most basic problems. Two basic questions that remain unanswered are: What proportion of molecular polymorphism, if any, is maintained by selection, and how? And, how important are the fitness effects of interactions among sequence residues? I review recent progress in answering these questions and present results from my own work on HIV-1.

T57. Population genetics and adaptation to drought in Eucalyptus camaldulensis

Rachel L. McEvoy¹, Gavin N. Rees², Shannon K. Dillon³, Yvonne M. Parsons¹

Department of Genetics, La Trobe University, Bundoora ²Murray-Darling Freshwater Research

Centre and CRC for Freshwater Ecology, Wodong ³CSIRO Plant Industry, Canberra

Australia has been experiencing one of the worst droughts on record for the past 10 years and in order to survive organisms need to adapt rapidly to these environmental changes. *Eucalyptus camaldulensis* is the most widespread eucalypt species, but inhabits watercourses and floodplains which may make it sensitive to the effects of drought. The aim of this study was to investigate adaptation to drought stress in *E. camaldulensis* in Yanga National Park, NSW, by determining levels of diversity, population structure and evidence of selection using SNP data from 12 drought tolerance candidate genes. Results indicate that drought stress due to different flood histories has a negative impact on tree health and survival, indicating that a strong selective pressure exists. Comparisons with 20 Australia-wide populations show that genetic structure is present in *E. camaldulensis*, especially at the regional level, which is correlated with distance rather than rainfall. The Yanga population is very similar to other Murray-Darling Basin populations, which differ greatly from all other regions. Our results also suggest that here is some evidence of selection in water and stress related genes within Yanga and Australia-wide.

T58. Disparate patterns of population genetic diversity, structure and clonality in a critically endangered *Banksia* species.

Melissa A Millar, Margaret Byrne, David Coates Science Division, Department of Environment and Conservation, Perth

Estimates of genetic diversity, genetic differentiation and degree of clonal reproduction can be used as criteria for assessing species conservation status and are also vital for planning appropriate conservation management actions. This is especially true for rare and endangered species which are more likely to employ a level of clonal reproduction. Multilocus genotyping in the critically endangered Western Australian Banksia ionthocarpa revealed predominantly clonal reproduction in populations of subspecies chrysophoenix, and no evidence of clonality in populations of subspecies ionthocarpa. Genotypic diversity is low in subspecies chrysophoenix, with a total of sixteen unique multilocus genotypes detected across four extant populations. Two populations are comprised of single genets, and in multiclonal populations dominant clonal patches cover large areas. The results indicate that current census sizes greatly overestimate the number of unique individuals of this taxon. Whilst overall genetic diversity is low in B. ionthocarpa compared to a number of other common Banksia species, subspecies chrysophoenix is also genetically depauperate compared to its sister taxon. highlights the utility of population genetic studies in assessing the genetic vulnerability of rare and endangered taxa, including those suspected of clonal reproduction, and provides recommendations for the appropriate conservation management of each subspecies.

T59. Assessing diversity of the MHC in the domestic cat, cheetah and Gir lion using MHC linked microsatellite markers

Katrina Morris¹, Julia Beatty¹, Sonia Cattley², Marilyn Menotti-Raymond³, Stephen O'Brien³, Katherine Belov¹

¹Faculty of Veterinary Science, The University of Sydney, NSW. ²Sydney Bioinformatics, The University of Sydney, NSW. ³Laboratory of Genomic Diversity, National Cancer Institute, Frederick, MD, USA.

The Major Histocompatibility Complex (MHC) encodes genes with an important role in immunity. Microsatellites linked to MHC genes act as proxies for measuring diversity in the peptide binding region of MHC genes by the hitchhiking effect. These microsatellites provide a means for studying the diversity of MHC genes and how they relate to disease and behaviour. We have developed six polymorphic microsatellite markers linked to both class I and class II MHC genes in the domestic cat (*Felis catus*) and used these to assess the diversity of the MHC in outbred domestic cats, Burmese cats, cheetahs (*Acinonyx jubatus*) and Gir lions (*Panthera leo persica*). Burmese cats show less MHC diversity than outbred domestic cats, though both populations have a heterozygote deficiency. The markers are polymorphic in the cheetah, with diversity comparable to that of the domestic cat, revealing a higher level of diversity in the cheetah MHC than previously estimated. All of the markers were monomorphic in the Gir lion supporting the hypothesis that the Gir lion has critically low genetic diversity.

T60. Investigating the presence of a cryptic specices-complex in *Apiomorpha minor* (Hemiptera: Sternorrhyncha: Coccoidea)

Penelope J. Mills
School of Biological of Sciences, UQ, St. Lucia.

Almost all species of scale insects have been described solely on the morphology of adult females. In the gall-inducing genus *Apiomorpha*, there is considerable variation in chromosome number, including diploid counts between 8 and 84 in a single described morphospecies (*A. minor*). This suggests that cryptic species might exist. The aim of this research was to use DNA sequence data to assess genetic variation in *A. minor* and determine whether cryptic species are present. We found that there are at least six cryptic species in the A. minor species-complex, which show some association with host-use. There was little congruence between chromosome number and genetic groupings.

T61. Paternity analysis of the autotetraploid invasive tree Salix cinerea in south-eastern Australia.

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Tara Hopley^{1,2}, Andrew Young¹

¹ CSIRO Plant Industry, Canberra, ACT, Australia ² Australian National University, School of Biology, Acton, ACT, Australia.

Paternity analysis in wild populations of plants is difficult due to: large potential paternal pools, extensive pollen dispersal distances, significant variation in reproductive effort among males and the complications introduced by polyploidy. Here we used newly developed paternity analysis procedures that explicitly take these factors into account to undertake an SSR-based assessment of pollen-mediated gene flow within and between populations of the autotetraploid tree *Salix cinerea*, an aggressive exotic component of river systems in southeastern Australia. Preliminary exclusion-based analysis using 6 loci show that up to 20% of seed on trees are sired from outside their home population. Genetic profiling of populations in surrounding rivers will allow us to identify the most likely pollen sources providing data on the scale of pollen movement. Parentage analysis to ascertain the origin of seedlings in these same populations will allow us to directly measure the scale of seed dispersal. These results will assist land managers to develop more effective eradication strategies by: 1) identifying the most efficient geographical scale of control that will minimise the likelihood of reinfestation; 2) identifying important source populations of willow propagules; 3) providing quantitative analysis of catchment that are most at risk of invasion in the future.

T62. Genome-wide

<u>Jeremy McRae</u>, Yael Salzman, Sara Jaeger, Richard Newcomb

The New Zealand Institute for Plant & Food Research Limited, Mt Albert, Auckland, New Zealand

Odorant receptors are the largest gene family in the human genome. Approximately 390 functional receptors discriminate over 10,000 odours, yet the ability to detect only a few odours has been traced back to specific receptors. The ability to perceive specific odours varies widely among individuals and is thought to be, at least in part, genetically determined.

We have been investigating variation in the ability to detect important flavour compounds in fruit. Extensive variation was detected in the ability to detect five compounds, with ß-ionone displaying variation across six orders of magnitude. Participants in the study were genotyped using Affymetrix SNP 6.0 microarrays. For the flavour compound *cis*-3-hexen-1-ol, which forms part of the 'grassy' odour of some fruit such as kiwifruit, we identified a cluster of significantly associated SNPs. Furthermore, this cluster of SNPs is co-localised among a cluster of genes encoding odor nt receptors.

T63. The immune system and insect sociality

Ross H Crozier, Helge Schlüns, Y Ching Crozier School of Marine & Tropical Biology, and Comparative Genomics Centre, JCU, Townsville

Social insects live in often densely-packed societies of genetically similar individuals. Herd immunity is expected to be important in improving group resistance to infection, and indeed parasite prevalence is negatively correlated with intracolony genetic diversity in ants. Consistent with this, experimental groups with more genetic diversity show higher resistance to fungal infection than such groups with less diversity. The rate of positive selection is also expected to vary with changes in habitat and social variables. We have found such variation in termite GNBPs and Relish. We now extend this to *Myrmecia* ant GNBPs and, due to the low detection capabilities of traditional dN/dS ratios for detecting positive selection, use physicochemical properties, which enable distinction between positive selection that is stabilizing and that which is destabilizing of protein attributes. Ant GNBPs showed significant positive selection for 16 amino acid properties of 31 investigated, with stabilizing selection indicated for 14, moderate destabilizing for one, and strong destabilizing selection for pK'. Termite GNBPs show similar but weaker tendencies, but termite Relish does not show strong destabilizing positive selection for pK'.

T64. Three's a crowd:- Wolbachia and pathogen resistance in insects

<u>Jeremy C. Brownlie¹</u>, Lauren M. Hedges², Scott L. O'Neill² and Karyn N. Johnson².

¹ School of Biomolecular and Physical Sciences. Griffith University, Brisbane. ² School of Biological Sciences. University of Queensland, Brisbane.

Wolbachia are maternally inherited bacterial symbionts that infect millions of different insect, spider and filarial nematode species and live exclusively inside the host cell — yet they have dramatic effects on host biology. Most research to date has focused on Wolbachia's unique ability to manipulate host sex determination or reproductive systems, however an increasing body of work has emerged that shows Wolbachia can provide a range of fitness benefits to their hosts. As Wolbachia form systemic and persistent infections for the lifetime of their insect host, Wolbachia are likely to come into contact with transient pathogens, such as viruses or fungi, which infect the same insect host. Generally, infection by a pathogen reduces the host's lifespan and reproduction — so how should a maternally inherited endosymbiont that depends upon its host to reproduce respond? Here I will discuss recent findings that shows Wolbachia can protect their hosts from insect pathogens and what impact this could have for both insect host and Wolbachia.

T65. A new set of Wolbachia-associated host phenotypes in the mosquito, Aedes aegypti

Elizabeth A. McGraw, Luciano Moreira, Andrew Turley, Scott O'Neill The School of Biological Sciences, The University of Queensland, St. Lucia, Qld, Australia 4072

It has been proposed that *Wolbachia* infections that limit mosquito lifespan might be used to reduce dengue transmission by *Aedes aegypti* since only old mosquitoes are capable of transmitting the virus. The successful introduction of a life-shortening strain of *Wolbachia* into *A. aegypti* has recently been reported that halves adult lifespan under laboratory conditions. In the infected mosquito we have also identified two additional *Wolbachia*-associated host phenotypes. First, as *Wolbachia* infected mosquitoes age, their ability to successfully obtain a blood meal decreases. This reduction appears to be caused by a failure of the proboscis to function. Second, the presence of *Wolbachia* infection reduces dengue virus load in mosquitoes. These findings reveal a broader role for *Wolbachia* in host physiology and together in combination with life-shortening offer potentially more powerful prospects for dengue control.

T66. Improvements to Transcriptional Profiling-based Age grading using in *Wolbachia*-infected *Aedes aegypti*

Eric Caragata, Luciano Moreira, Scott O'Neill, Elizabeth McGraw School of Biological Sciences, The University of Queensland, St. Lucia, Qld 4072, Australia

The development of a precise method of predicting mosquito age is required to accurately evaluate changes to mosquito population age-structure resulting from the introgression of Wolbachia-infected Aedes aegypti mosquitoes. Currently the most accurate method of age grading for mosquitoes utilises age-dependent changes in a set of gene transcripts. Unfortunately, age predictions made using this model decrease in accuracy for older, epidemiologically important mosquitoes. This decrease is a product of non-performing genes that show little change of expression with age. We conducted a microarray assay to identify novel candidate genes that showed significant changes in expression with age. The transcripts of two genes AAEL007490 and AAEL014255 showed significant and consistent decrease in expression with age. qRT-PCR analysis of these transcripts and the two best performing transcripts from those previously identified, produced an age-grading model with a 3% stronger relationship with age for wMelPop-infected mosquitoes, and 5% stronger for uninfected mosquitoes. All four transcripts showed similar patterns of expression regardless of expression status. Age predictions conducted using these models showed an average increase in accuracy of 1.5 days for infected and 2 days for uninfected mosquitoes. Predictions made using both models maintained similar levels of accuracy out to 29 days of age.

T67. Male-biased survival to infection and immune gene expression in *Drosophila* melanogaster

Yixin H. Ye, Elizabeth A. McGraw School of Biological Sciences, The University of Queensland, St. Lucia, Qld 4072, Australia

Sex differences in immune functions are common and females usually produce a more robust immune response and are less susceptible to infection. This has been well-documented in humans, mice, certain species of birds and reptiles. A variety of hypotheses have been put forward which usually propose that males gain fitness by increasing their mating success at the cost of immune investment whilst females invest heavily in reproduction. As a consequence females are also predicted to invest more in immunity to maximize life time egg production. In insects, however, there are contradicting evidences of a male-biased investment in immunity. In a population of D. melanogaster derived from the wild we selected for improved defense to systemic infection with the opportunistic pathogen Pseudomonas aeruginosa and found male flies consistently exhibits a higher survival rate post-infection compared to females. We then picked a set of immune genes whose expression had previously been shown to confer defense against P. aeruginosa and studied of their expression in both sexes at the basal and the induced levels. We found that male-biased survival to infection is at least partially attributable to a higher baseline expression of immune genes in males. Contrary to previous published knowledge about host response to P. aeruginosa infection, we found the immune genes are readily induced after infection, however the two sexes respond in the same magnitude in immune gene induction after infection. Lastly, we found that selection only alters the expression of the genes at the induced state and males appear to have a stronger response to selection than females. Together our findings suggest a superior immune response in male Drosophila.

T68. Virus protection in Drosophila melanogaster by Wolbachia

<u>Lauren M. Hedges</u>¹, Jeremy C. Brownlie², Scott L. O'Neill¹, Karyn N. Johnson¹

¹School of Biological Sciences, UQ, Brisbane. ²School of Biomolecular and Physical Sciences, Griffith University, Brisbane.

Host-pathogen interactions are complex, involving pathogen mechanisms of infection and host immune responses. Drosophila C virus is a simple, well studied virus which is highly pathogenic to *Drosophila melanogaster*, and this system is a useful model for studying virus: host interactions in insects. DCV is a non-enveloped virus with a single-stranded positive-sense RNA genome, belonging to the *Dicistroviridae* family. Previous studies have identified *Drosophila* genes that are specifically up-regulated in response to virus infection. While investigating the up-regulation of three of these genes by DCV subvirus components, a fly line was found where these virus induced genes were not up-regulated as expected. In addition, mortality caused by virus infection was delayed in this line. This *Drosophila* line was found to be infected with *Wolbachia*, a gram-negative, obligate, intracellular bacteria which is maternally transmitted. A series of experiments confirmed that the presence of *Wolbachia* affects virus: host interactions in this model, decreasing DCV accumulation and delaying *Drosophila* mortality.

T69. Genomes and sex chromosomes of Australia's weird animals – how my Honours project became my life's work

Jennifer Graves

ARC Centre of Kangaroo Genomics, Research School of Biology, The Australian National University, Canberra ACT 2601, Australia.

When I left Adelaide for a PhD in Berkeley in 1965, I thought I had left marsupial chromosomes behind in David Hayman's lab, where I did my Honours on X inactivation in kangaroos. Indeed, if it were not for Des Cooper's cajoling when I returned to Australia and started at La Trobe, I would have gone on with cell cycle control studies in mutant mouse cells. I recall rudely rejecting Des' appeal to use my cell fusion expertise to map genes on kangaroo X chromosomes, saying, "why would anyone want to do *that*?" David and Des appreciated, much better than I, that comparing the genes and chromosomes of distantly related mammals would offer new insights into fundamental processes of chromosome function, regulation and evolution.

And so they have. Not just kangaroos, but dunnarts and platypus, devils (Tasmanian) and dragons (lizards). Australia is crawling (bounding) with unique fauna that, like mutant mice, provide genetic variation to study the fundamental biological processes. I have capitalized on this variation to examine how mammal genomes evolved, how gene families evolved and how they work. In particular, I use them to study the peculiarities of sex chromosomes; their function, epigenetic modification and how they evolved from an autosome that is still extant in platypus.

First we slowly and painfully built maps of the kangaroo X, which we used to look at X chromosome inactivation. I joined the Comparative Mapping committee of the Human Gene Mapping workshops, the fore-runner of the Human Genome Project. Initially an outsider in this mousy committee, I enthused about kangaroos to audiences that laughed, although they did appreciate that kangaroo mapping revealed the evolutionary layers of human sex chromosomes. Then a surprise phone call, and suddenly we were working on sex determination. Our finding that the then candidate sex determining gene was on an autosome, not the Y in kangaroos exposed it as the wrong gene; my student later cloned the right gene and we discovered a gene on the X from which it evolved. Not only did this discovery catapult my group into the thick of the sex wars, but it showed how marsupials could deliver unique information. The same thing has happened again and again; we accidentally discovered new human mental retardation, globin, and testis genes, and we overturned accepted theories about the age and fate of the Y chromosome and the mechanism of X inactivation. Nobody laughs any more.

As everyone (except nasty referees) knows, doing genetics on non-model mammals is tough. For years, we stretched cytogenetic methods to map kangaroo and platypus genes, and scratched around trying to put together tiny pieces of cloned genes. The complete genome sequencing of the opossum, kangaroo and platypus now provides avalanches of data on marsupials and monotremes to anyone in the world, so I hope that ever more Australian geneticists will use data from our own animals to answer some of the big questions in biology.

Abstracts (Poster presentations):

P1. Use of evolutionary conservation to study the role of Vps22: a key endosomal protein

Jeffrinder Singh, Renzo D'Ortenzio, Jasmina Ilievska, <u>Naomi Bishop</u>
Cell Biology and Molecular Pathogenesis Laboratory, Department of Microbiology, La Trobe
University, Bundoora VIC

The Endosomal Sorting Complex Required for Transport (ESCRT) machinery is essential for the biogenesis of MultiVesicular Bodies (MVB), organelles with a pivotal role in the endosomal pathway. In metazoan and fungal cells four ESCRT complexes (ESCRT0-III) are recruited sequentially to the endosomal membrane. An ATPase, Vps4 then plays a role in dissociation of the ESCRT complexes from membranes and in fission of intraluminal vesicles within MVBs. The ESCRT complexes play a crucial role in receptor down-regulation, retroviral budding and cell division. The ESCRTIIII–Vps4 interaction predates the divergence of Archaea and Eukarya and functions in archael cell division. ESCRTI-III are present in all major eukaryotic supergroups, but some clades have lost genes encoding ESCRTI and/or ESCRTII components. ESCRT-0, by contrast, is only found in fungi and metazoan genomes. ESCRTII consists of two copies of Vps25, and one copy each of Vps22 and Vps36. We have studied the evolutionary history of Vps22 in eukaryotes and report on the evolutionary conservation of Vps22, the conservation of Vps22 tertiary and secondary structural elements, and identify novel motifs within Vps22, which we suggest have a key role in ESCRTII function.

P2. Genetic variation of MHC class I in two Collared peccary (Pecari tajacu) populations

Amanda Y. Chong¹, Cibele Biondo², Jaime Gongora¹

¹Centre for Advanced Technologies in Animal Genetics and Reproduction, Faculty of Veterinary Science, University of Sydney, NSW 2006, Australia. ²Departamento de Ecologia, Instituto de Biociências, Universidade Estadual Paulista UNESP-Rio Claro, Brazil

The Collared peccary (*Pecari tajacu*) from the Americas is a distant relative of the Eurasian pig. Due to a growing interest in its use for commercial farming, it offers a unique opportunity to study the effects of captivity and selection for commercial traits on immune function. Despite some attempts to characterise some genes of this genomic region in peccaries using the porcine Major Histocompatibility Complex (MHC) resources, studies within the Collared peccary have never been attempted. Here we present preliminary data on genetic variation in the antigen-binding region of an MHC class I gene within two farmed and wild Collared peccary populations from Brazil. Preliminary analyses show that between one and two alleles were found per individual suggesting that a single locus is being amplified. Data collected during this study could provide an insight into the selection pressures imposed on captive populations with relevance to peccary diseases, conservation and farming.

P3. Microevolution of *Cryptococcus neoformans* driven by massive tandem gene amplification

<u>Eve W. L. Chow</u>, Alan W. L. Law, James A. Fraser *University of Queensland*

In various organisms, subtelomeric regions of the chromosomes have been shown to undergo a much higher rate of rearrangement. These changes often take the form of gene amplification events. We hypothesised that similar genetic changes occur in the subtelomeres of the human pathogen *Cryptococcus neoformans*, playing a role in the persistence of infection. While characterising these changes, we identified a gene amplification event in a number of clinical isolates involving an arsenite-efflux transporter. The 3,177 bp *ARR3* gene amplicon exists in tandem arrays of 1 – 15 copies with MLST studies revealing that strains bearing this naturally occurring amplification belong to *C. neoformans* var. *grubii* VNI subclade A5; these strains display enhanced but variable resistance to arsenite and arsenate that correlates with copy number of the repeat. The arsenite sensitivity of the *S. cerevisiae arr3* mutant is complemented with the *C. neoformans ARR3* gene. Microevolution experiments have shown that further massive amplification is easily induced through passage on increasing concentrations of arsenite (up to 30 mM), yielding gene amplifications that account for ~1% of the genome. This finding is a proof of concept that adaption through gene amplification may be one mechanism that *C. neoformans* employs during infection.

P4. Levels of heat tolerance in *Drosophila melanogaster* may depend on upstream regulation of the heat shock response

<u>Fiona E. Cockerell</u>, Travis K. Johnson, Carla Sgro, Stephen W. McKechnie Centre for Environmental Stress and Adaptation Research (CESAR), School of Biological Sciences, Monash University

Heat tolerance is an important adaptive trait since it varies across diverse *Drosophila* species in a manner consistent with their diverse altitudinal and latitudinal habitats. Heat tolerance also varies within species according to local temperature conditions, such as the latitudinal heat tolerance cline in *D. melanogaster* along the Australian east coast. While genotype variation in some genes of the cellular heat stress response has been associated with heat tolerance in this species the effects of variation in expression of these genes has received less attention. Here we characterise expression levels of Hsp70, the major and well-characterised inducible Hsp, and Hsp83 in a set of 40 isofemale lines of *D. melanogaster* derived from a natural population and look for associations with heat tolerance. *Hsp83* (sometimes referred to as Hsp90) is ubiquitously expressed in all organisms under non-stress conditions and is modestly upregulated upon heat stress. Our results suggest that Hsp83 expression, but not Hsp70, is related to heat tolerance and that changes in the regulation of the cellular heat shock response may be the important determinant of heat tolerance variation in this species.

P5. Conservation of motifs and domains in endosomal sorting protein Vps36

Renzo D'Ortenzio, Jeffrinder Singh, Jasmina Ilievska, Naomi Bishop

Cell Biology and Molecular Pathogenesis Laboratory, Department of Microbiology, La Trobe University, Bundoora VIC

Split pleckstrin homology (PH) domains are a subclass of PH domains characterized by the insertion of an autonomously folded protein module(s) within the PH domain sequence. Split PH domains are found in various proteins, including syntrophins, Rho-family GTPases, and the Vps36 subunit of the yeast ESCRT-II complex; the latter being a complex playing an important role in eukaryotic endosomal trafficking. Yeast Vps36 has an insertion of two NZF (Np14 zinc finger) motifs within its N-terminally located PH domain, one of which can bind ubiquitin, an interaction required for Vps36 endosomal function. By contrast, the mammalian orthologue of Vps36, Eap45, has an uninterrupted PH domain, known as the GLUE domain (GRAM-like ubiquitin-binding in Eap45). Unexpectedly, the Eap45 GLUE domain retains the ability to bind ubiquitin. We have studied the evolution of the Vps36 family, including the evolutionary history of the acquisition of the tandem NZF motifs in yeast. Our analyses also provide additional information on the evolutionary conservation of *Vps36* across the eukaryotic super-groups, the conservation of Vps36 tertiary and secondary structural elements, and identify novel motifs that may play a key role in Vps36 endosomal function.

P6. A genome-wide scan reveals candidate loci associated with differential viability between the sexes in humans

<u>Jinghua Feng</u>, Simon Easteal, Gavin A. Huttley

The John Curtin School of Medical Research, ANU, Canberra

In the human population, the observation of significant discordance of genotype distributions in males and females from the same generations can be explained by differential viability between the sexes, when mutation is omitted. The extent to which this pattern of natural selection is shaping the human genome is unknown. Here we will show the results for comparing the genotype distributions of the two sexes at 500,568 single-nucleotide polymorphisms (SNPs) in 16,179 subjects of Caucasian ancestry (7,711 males and 8,468 females), and show the candidate loci with the strongest signal which are associated with differential viability between the sexes.

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P7. Phylogeographic analyses of platypuses from continental mainland and Tasmania using mitochondrial DNA

Amelia Swan¹, Stephen Kolomyjec², Elise Furlan³, Tom Grant⁴, David Blair², Chris Moran¹, Andrew Weeks³, Nick Gustand⁵, <u>Jaime Gongora¹</u>

¹Centre for Advanced Technologies in Animal Genetics and Reproduction, Faculty of Veterinary Science, RMC Gunn Building B19. University of Sydney, NSW 200, ² School of Marine and Tropical Biology, James Cook University, QLD 4811. ³ Bio21 Institute, University of Melbourne, VIC 3010, ⁴School of Biological, Earth and Environmental Sciences, University of New South Wales, NSW 2052. ⁵ Biodiversity Conservation Branch, Department of Primary Industries and Water, TAS 7001.

The platypus (*Ornithorhynchus anatinus*) is a monotreme endemic to Australia. Its current distribution is the streams and rivers of southern and eastern Australia and Tasmania. The platypus as a representative of monotremes has been used as a model for comparative genomic analyses providing interesting insights into the evolution of vertebrates. Besides these important contributions, understanding of the genetic diversity and phylogeography of the species is very limited. Here we analyse the phylogenetic relationships of platypuses from New South Wales, Australian Capital Territory, Queensland, Victoria and Tasmania using mitochondrial DNA control region sequences. Preliminary analyses show that platypuses from Tasmania cluster in a separate clade from those of continental origin. Within continental Australia, the phylogeographic structure appears to be relatively weak. To further understand the phylogeographic relationships of this species, more than 300 samples are being analysed. This information, in addition to current microsatellite studies will contribute to the assessment of the level of genetic differentiation and population structure of the platypus, assisting molecular ecology studies and conservation plans.

P8. Insights into the Major Histocompatibility Complex of crocodilians

Weerachai Jaratlerdsiri¹, Sally R. Isberg^{1,2}, Jaime Gongora¹

¹Faculty of Veterinary Science, University of Sydney, NSW, 2006, Australia. ²Porosus Pty Ltd, PO Box 86, Palmerston NT 0831, Australia

Crocodilians (alligators, crocodiles and gharials) are important for the wildlife tourism and emerging animal industries across the world. As a result of captive management, crocodilians have been faced with health challenges. It is considered that characterisation of the Major Histocompatibility Complex (MHC) in this lineage will be central to understand the immune response to these challenges. Here, we present preliminary analyses of MHC class I and II genes from the Australian saltwater crocodile and assess the diversity of one of these genes among twenty extant species of crocodilians. Comparison of predicted amino acid sequences show that the highest affinity is found between saltwater crocodile and reptile MHC class I genes (72%) while class II gene shows equivalent levels (~41%) of similarity with reptiles, avian and placental mammals. MHC class II β (exon 2) sequences show 91% similarity among crocodilian species and between one and three alleles per individual suggesting that more than one locus is being amplified for some species. This project is an essential first step in further studies to assess the mechanisms behind disease emergence in wild and farmed crocodilians and to understand the loss and/or gain of some genes during the diversification of this lineage.

P9. Nitrogen metabolite repression in *Cryptococcus neoformans*: A global regulatory network linking nitrogen scavenging and virulence factors

<u>Ivor R. Lee</u>, James A. Fraser *University of Queensland*

The ecological niche of the basidiomycete human pathogen *Cryptococcus neoformans* is nitrogen-rich pigeon guano. While nitrogen acquisition and metabolism is essential for microbial growth, it can also contribute to pathogenicity.

Nitrogen metabolite repression is a well-studied regulatory phenomenon in fungi belonging to the phylum *Ascomycota* in which it is mediated by GATA transcription factors. We hypothesise that one or more GATA factors may be the key regulator of permeases and catabolic enzymes associated with nitrogen metabolism in *C. neoformans*. Here we provide the first evidence of nitrogen metabolite repression in a member of the phylum *Basidiomycota*, revealing that beyond the traditional role in nitrogen scavenging it is a key regulatory mechanism controlling the coordinated expression of virulence factors such as the metalloenzyme urease and the polysaccharide capsule.

In an effort to identify the transcription factor mediating this effect, we created deletion mutants for several predicted GATA-type genes. We identified one such gene, *ARE1*, essential for the utilisation of various nitrogen sources including ammonium, a preferred nitrogen source in fungi, and uric acid, the predominant constituent of nitrogen in pigeon guano. Unexpectedly, Are1 also functions to negatively regulate growth at human body temperature. These studies suggest that beyond controlling the regulon of genes for nitrogen acquisition, Are1 is a key regulator of virulence-associated phenotypes.

P10. Characterisation of MHC-linked microsatellite markers in the platypus (*Ornithorhynchus anatinus*)

Lillie, M.¹, Gust, N.², Belov, K.¹

¹Faculty of Veterinary Science, University of Sydney ²Tasmanian Department of Primary Industries and Water

The platypus (*Omithorhynchus anatinus*) faces an uncertain future from the impact of a variety of potential threats, including climate change, human encroachment causing habitat degradation and the infectious disease, mucormycosis. In this project, we will apply measurements of genetic diversity in Major Histocompatibility Complex (MHC) genes, which are linked with innate and adaptive immune responses, to evaluate the immunological fitness and evolutionary potential of platypus populations. Microsatellite markers located in close proximity to MHC loci presents a simple, inexpensive and high throughput method of inferring genetic diversity at the MHC due to tight linkage with specific MHC alleles. This project aims to develop such microsatellite markers adjacent to MHC Class I and Class II loci to facilitate rapid typing of MHC diversity in platypus populations sampled from Queensland, New South Wales, Victoria, Tasmania, King Island and Kangaroo Island. Preliminary results indicate monomorphy in the King Island populations at two different MHC-linked microsatellite markers, indicating that this small island population is vulnerable to disease epidemics. Unique alleles have been identified in the Tasmanian population, increasing support for its sub-population classification. Ultimately, this study will assist conservation efforts.

P11. Towards the generation of induced Pluripotent Stem (iPS) cells from non-eutherian mammals

<u>loannis J Limnios¹</u>, Klaus I Matthaei², Margaret L Delbridge^{1,3}, Jennifer A Marshall Graves^{1,3}, Ernst J Wolvetang⁴.

¹Research School of Biology, ANU, Canberra, ²John Curtin School of Medical Research, ANU, Canberra, ³ARC Centre of Excellence for Kangaroo Genomics, Research School of Biology, ANU, Canberra, ⁴Australian Institute of Bioengineering and Nanotechnology, University of Queensland, Brisbane.

The molecular regulation of pluripotency in the early embryo of placental mammals has been primarily studied using mouse and human embryonic stem (ES) cells. Despite several efforts, no marsupial ES cell lines have been established, and to our knowledge there have been no attempts to isolate monotreme ES cells. Induced Pluripotent Stem (iPS) cells provide a functional alternative to obtaining ES cells through the reprogramming of differentiated somatic cells back to an embryonic-like state, as demonstrated in mouse and human. The aim of this project is to explore mechanisms to generate marsupial and monotreme iPS cells. To initiate this project we are looking at the effects of over-expressing the human reprogramming factors (Oct4, Sox2, Nanog, Lin28, cMyc and Klf4) in human, marsupial and monotreme fibroblasts. We are trialing several reprogramming methods, including use of lentivirus, episomes and protein extracts. This poster highlights the rationale, strategy and preliminary data from the project to date. If successful, marsupial and monotreme iPS cells will provide powerful *in vitro* models to study both the evolution of mammalian pluripotency and to test hypotheses to account for genomic and epigenetic phenomena.

P12. DNA-based identification, phylogeny and thermobiology of the forensically important Australian Sarcophagidae (Diptera)

Post of award

<u>Kelly A. Meiklejohn¹</u>, James F. Wallman¹, Mark Dowton²

¹Institute for Conservation Biology and Law, School of Biological Sciences, University of Wollongong, Wollongong. ² Institute for Biomolecular Science, School of Biological Sciences, University of Wollongong, Wollongong.

The utility of the forensically important Sarcophagidae (Diptera) for time since death estimates has been severely limited, as morphological identification is difficult and thermobiological histories are inadequately documented. A molecular based identification method, involving the sequencing of the COI 'barcode' fragment from 85 adult specimens, representing 16 Australian species from varying populations, was evaluated. The effectiveness was assessed by calculating the nucleotide sequence divergences using the Kimura-two-parameter (K2P) distance model, and generating neighbour-joining (NJ) phylogenetic trees. All species were resolved as reciprocally monophyletic, except Sarcophaga dux. Intraspecific and interspecific variation ranged from 0.000-1.499% (SE = 0.044%) and 6.658-8.983% (SE = 0.653%), respectively. Three maximum likelihood trees (based on different outgroups) were generated to examine the preliminary phylogeny of the Australian Sarcophagidae based on the COI 'barcode' sequence data set. Clear evolutionary relationships for the Australian Sarcophagidae species were not robustly obtained. Preliminary thermobiological studies were also conducted on the endemic and forensically important Australian sarcophagid, Sarcophaga impatiens. This showed that S. impatiens required 432 hours for development from larva to adult under the common development temperature (~25°C), with 120 hours spent in the larval stages (first, second and third instar) and the remaining as pupae.

P13. The olfactory receptor gene family of tammar wallaby (Macropus eugenii)

Amir Mohammadi, Hardip R. Patel, Margaret L. Delbridge, Jennifer A.M. Graves ARC Centre of Excellence for Kangaroo Genomics, Research School of Biological Sciences, ANU, Canberra.

Olfactory receptors (ORs) are seven-transmembrane-domain, G protein-coupled proteins that are responsible for binding odorants in the nasal epithelium. Olfactory receptor genes constitute the largest mammalian gene family with more than 1000 genes in some species. We have identified the OR gene repertoire from the first genome assembly of the tammar wallaby. 1511 opossum (*Monodelphis domestica*) OR protein sequences were used as query in a reciprocal TBLASTN search and 1457 sequences were isolated representing 1409 unique tammar wallaby OR genes. Therefore there are almost similar numbers of OR genes in both marsupial species. In addition, like the OR gene family in opossum, it is estimated that almost half of the tammar ORs are pseudogenized by one or more frameshift and/or insertion/deletion mutations. Tammar wallaby OR gene sequences were conceptually translated and were aligned with OR protein sequences from opossum, platypus, dog, rat, mouse and human. We have constructed phylogenetic tree for all OR genes from seven extant mammalian species and classified tammar wallaby OR genes into families and subfamilies. The OR genes were bioinformatically mapped to tammar wallaby chromosomes and further analysis has revealed some marsupial specific features in the evolution of this gene family.

P14. GTP biosynthesis and antifungal drug targets in Cryptococcus

Carl A. Morrow, Ulrike Kappler, Anna Stamp, Eugene Valkov, Justine M. Hill, Bostjan Kobe, James A. Fraser School of Chemistry and Molecular Biosciences, The University of Queensland, St Lucia, Queensland, 4072 Australia

Novel therapeutics and synergistic approaches to treatment are required for opportunistic fungal infections due to the increasing incidence of relapse and resistance among the AIDS population combined with the lack of effective antifungal agents. We have investigated the GTP biosynthesis and salvage pathways in the human pathogenic fungus *Cryptococcus neoformans*, a common cause of fatal fungal meningoencephalitis in primarily immunocompromised patients worldwide. We have identified and characterised homologues of *IMD1*, encoding the rate-limiting enzyme inosine 5'-monophosphate dehydrogenase and *HPT1*, encoding the purine salvage enzyme hypoxanthine xanthine guanine phosphoribosyltransferase, via targeted gene disruptions and phenotypic assays. Additionally, we have examined the basis for naturally occurring susceptibility and resistance to the IMP dehydrogenase inhibitor mycophenolic acid through the creation of mutant and chimaeric *IMD1* alleles. Detailed structural and functional analysis of two *Cryptococcus* IMP dehydrogenase alleles reveals further insight into the mechanism of this potential antifungal drug target.

P15. Population structure of South Asian indigenous pigs determined by microsatellite markers

¹Karma Nidup^{1,2}, G.L.L.Pradeepa Silva³, D.D.Joshi⁴, Rinzin Pem⁵, Jaime Gongora¹, Chris Moran¹

¹Centre for Advanced Technologies in Animal Genetics and Reproduction (REPROGEN), Faculty of Veterinary Science, University of Sydney, Australia. ²College of Natural Resources, Royal University of Bhutan, Lobesa, Punakha, Bhutan. ³Department of Animal Science, University of Peradeniya, Sri Lanka ⁴National Zoonoses and Food Hygiene Research Centre, Kathmandu, Nepal. ⁵Regional Veterinary Laboratory, Department of Livestock, Ministry of Agriculture, Gelephu,

Indigenous pigs ($Sus\ scrofa$) have socio-economic, cultural and traditional, and biodiversity importance in the lives of many people around the world including Bhutan, Nepal, and Sri Lanka (South Asia). Currently, there is very limited genetic information on South Asian pigs. Here, we investigate the genetic structure and diversity of both indigenous domestic and wild pigs (n=303) from South Asia as well as some Australian commercial pigs (n=15) of European origin using 21 microsatellites markers recommended by ISAG/FAO. Analysis of genetic structure reveals five different populations of pigs from Bhutan, two from Nepal, and clear segregation between village and wild pigs of Sri Lanka. Preliminary data indicates that country-wise samples deviated (P < 0.05) from HWE at most loci. The mean expected heterozygosity ranges from 0.70 to 0.81 (SE = 0.02) for Bhutanese pigs, 0.77 and 0.84 (SE = 0.01) for Nepalese pigs, 0.81 and 0.84 (SE = 0.01) for Sri Lankan pigs, and 0.67 (SE = 03) for Australian commercials pigs that are used as an out-group. Interestingly, Bhutanese and Nepalese pigs are more closely related when compared with Sri Lankan pigs. Our findings will be useful for conservation and sustainable utilization of porcine genetic resources in the region.

P16. Why can't we delete those deleterious Drosophila genes?

John Sved, Xiumei Liang School of Biological Sciences, Sydney University

Wild type chromosome homozygotes from natural *D. melanogaster* populations are only around 20% as fit as normal outbred flies. This is consistent with either widespread heterozygote advantage or with high levels of deleterious recessives. There seems little belief any more in the idea of widespread heterozygote advantage, so that leaves the deleterious recessives. At low frequency, there is little selective pressure on such genes. But at high frequency, 50%, there should be substantial disadvantage, leading to 'purging'. We previously conducted an experiment of this type, the di-chromosomal experiment, which failed to show evidence of purging. The experiment reported here, the di-parental experiment, avoids the possibility of dysgenic mutation, with deleterious genes raised to either 25% or 50% frequency. Computer simulation shows that the potential effect ought to be as great as in the di-chromosomal experiment. Once again we were unable to demonstrate purging. Any deleterious recessives must thus be of very small effect. But the result seems consistent with long-term inbreeding experiments of BDH Latter, and with inferences from the divergence of *D. melanogaster* and *D. simulans*.

P17. Lab:² A Pilot Study on Building Web Platform for Geneticists

Paul LF Tang, Carlos Lu Xiang, You-Qiang Song

Department of Biochemistry, Li Ka Shing Faculty of Medicine, The University of Hong Kong, Hong Kong China

The enhancement of web technology and internet infrastructure dramatically changed the way of our daily communication. In this Web 2.0 era, we can easily manage and share our photos, articles or even bookmark via a number of online services (e.g. Flickr, Facebook), contribute our knowledge to the public domain (e.g. Wikipedia, Freebase) or even create a virtual world (e.g. SecondLife, Google Lively). The internet becomes a square for data generation, transfer and discussion.

In a laboratory, with the terabytes of information being generated from different dimension of the biological systems from different sources with different technologies, data collection, integration and ultimately sharing had posted a major problem for scientific research. Mouse is one of the model organism with a huge amount of data being generated everyday from different laboratories. However, laboratory results are often kept in one's lab notebook and seldom aim for sharing. While the practice of having a lab notebook is indeed very good, however, the one dimension paper based recording method has become inadequate when compared to the speed and volume of current research. Recent efforts from mouse communities in sequencing 15 inbred strains, phenotyping more than 100 mouse strains on more than 100 phenotypes, cataloging embryo's gene expression pattern throughout the whole developmental stages and generating a set of collaborative cross of 1000 mouse lines from 8 inbred strains for potential mapping of disease-causing genes to 1Mb, post a serious problem for one to organize the huge amount of data and trigger the need of collaboration and sharing of laboratory results.

Inspired by the Open Science communities and recent advancement in web technologies and products, we initiate this project to explore ways of using different Web 2.0 services or technologies to facilitate geneticists to 1) record, organize and visualize the data generated from a laboratory in electronic form, 2) correlate one's own data with that in the public domain and 3) share with the public communities and ultimately and hopefully practice open science. With the aim of a ploit study, we focus on SNP (Single Nucletide Polymorphism) data, which is most common and with simplest data structure in genetics study. The experience gains here could be transferred to other data types (e.g. pedigree).

P18. Evidence that localised variation in primate sequence divergence arises from an influence of nucleosome placement on DNA repair

Hua Ying¹, Julian Epps ², Rohan Williams ¹, Gavin Huttley ¹

¹ John Curtin School of Medical Research, Australian National University, Canberra, ACT, Australia ² School of Electrical Engineering and Telecommunications, The University of New South Wales, Sydney, NSW Australia

Localised substitution rate heterogeneity exists in mammal genomes, but its origin remains unclear. Experimental studies established that chromatin compaction affects both DNA lesion formation and repair, which suggest that both the total substitution rate and types of substitution will be affected by chromatin status. The regular positioning of nucleosomes, the basic packaging unit of chromatin, further predict that substitution rate should vary spatially in an oscillating manner. We addressed the influence chromatin on substitution rate and types using recent genome-wide mapping of intronic and intergenic DNase I Hypersensitive sites (DHS) data. The flanking sites of DHS (Flank) were used as a closed chromatin matched control. For both intergenic and intronic samples, comparing matched DHS and Flank revealed an significant excesses of total substitution and transition substitutions in closed chromatin. We measured localised fluctuation in substitution rate along ~1800 primate promoters using phylogenetic footprinting. Using signal processing techniques, a dominant oscillation at ~200 bp was evident in both the substitution spectrum and nucleosome score derived from a Chip-seq assay of human T-cells. Our results support a role for differential DNA repair rates between open and closed chromatin in the spatial distribution of rate heterogeneity.

Delegate list:

Graves

Jenny

Shafagh Al Nadaf The Australian National University King Saud University Asama Al-hugail Sureshkumar Balasubramanian The University of Queensland Bill Ballard The University of New South Wales Victor Chang Cardiac Research Institute Ballouz Sara The University of New England & The University of Queensland Barker Stuart Philip Batterham The University of Melbourne The University of Sydney Madeleine Beekman The University of Sydney Kathy Belov The Australian National University Hannah Bender The University of Western Sydney Hawkesbury Melissa Berry La Trobe University Naomi Bishop Broadhurst **CSIRO Plant Industry** Linda **Griffith University** Jeremy Bronwnlie Landcare Research **Buckley** Thomas Richard Monash University Burke S Deborah Burnett The University of Sydney Walter and Eliza Hall Institute Rachel Burt Department of Environment and Conservation Margaret Byrne The University of Queensland Eric Caragata Flinders University David Catcheside The University of Queensland Steve Chenoweth The University of Sydney Amanda Chong The University of Queensland Chow Eve Cornell University Andrew Clark The University of Queensland Charles Claudianos **Fiona** Monash University Cockerell The University of Adelaide Alan Cooper James Cook University Ross Crozier The University of Adelaide Jack da Silva S Jonathan Davis Macquarie University The Australian National University Janine Deakin Macquarie University Jenny Donald La Trobe University Renzo D'Ortenzio The University of Wollongong Mark Downton The University of Sydney Alen Faiz The Australian National University Jinghua Feng Rod **Finney** Crown Scientific The University of Queensland **James** Fraser The University of Queensland Gibson Greg Primary Industries and Fisheries Rosie Godwin The University of Sydney Jaime Gongora

The Australian National University

Margaret Griffin Sarstedt Anita Grounder Crown Scientific Frank Grützner The University of Adelaide S Joanna Hare Macquarie University Lauren The University of Queensland Hedges Tara Hopley CSIRO/The Australian National University Gary Landcare Research, NZ Ltd Houliston Joel Griffith University Huev S Elizabeth The University of Adelaide Irvin Weerachai Jaratlerdsiri The University of Sydney Janine Jory Monash University Karin Kassahn The University of Queensland **Natalie** Judge Invitrogen The University of New England Margaret Katz Daphne Kusters Invitrogen Dave Lambert Griffith University Ivor Russel The University of Queensland Lee The University of Sydney Mette Lillie Jason Limnios The Australian National University The Australian National University Helen Lindsay Ali Livernois The Australian National University Andrew Lloyd The University of Adelaide S Hilary Martin The University of Queensland Rachel McEvoy La Trobe University McGraw Elizabeth The University of Queensland Steve McKechnie Monash University The New Zealand Institute for Plant & Food Research Jeremy McRae Kelly The University of Wollongong Meiklejohn Lee Miles The University of Sydney Department of Environment and Conservation Melissa Millar Penelope Mills The University of Queensland Amir Mohammadi The Australian National University Chris Moran The University of Sydney Katrina Morris The University of Sydney Carl Morrow The University of Queensland **Bridget** Murphy The University of Sydney Corina Murphy Genesearch Pty Veronica The Australian National University Murtagh Richard Newcomb Plant & Food Research Karma Nidup The University of Sydney Ben Oldroyd The University of Sydney Toni O'Keefe Crown Scientific Denis The Australian National University O'Meally Scott O'Neill The University of Queensland Ovenden Primary Industries and Fisheries Jenny Vidushi Patel The Australian National University Hardip **Patel** The Australian National University

Roche

Rast Student

Sarah

Peaty

Angelique Riepsamen The University of Wollongong Otto Schmidt The University of Adelaide The University of New South Wales Jeremy Shearman The University of Sydney John Sved The University of Hong Kong Paul Tang Sarstedt Trevor Taylor Telonis-Scott The University of Melbourne Marina S Wee The University of New South Wales Teo Timmis The University of Adelaide Jeremy Walter and Eliza Hall Institute Matthew Wakefield The University of Queensland Jenny Wang-Holmes Coral Warr Monash University The University of Sydney Peter Waterhouse The Australian National University Paul Waters \$ Cali The University of Sydney Willet The Australian National University Rohan Williams The University of New South Wales Alan Wilton The University of Sydney **Emily** Wong The University of Queensland Yixin Ye Genesearch Pty Bonni Yee Monash University Winston Yee Hua Ying The Australian National University **CSIRO** Andrew Young

Additions/Corrections to the abstract book:

Correction:

Greg Gibson's plenary will be held in building 8 room 139, not room 217.

Asma Al-Huqail (T53, session 8) will now be giving a poster instead of an oral presentation.

Recent additions to the delegate list:

First name	Surname	Institution
Katherine	Allen	Macquarie University
Michael	Arthur	Griffith University
Tim	Heupink	Griffith University
Holland	Kaplan	The Australian National University
Craig	Koina	Pacific Laboratory Products
Liz	Pearson	The University of New England
Sarah	Peaty	Roche

Poster presentations:

Please put your poster up by the first coffee break on the morning of the 8th. Judges of the student posters will want to see them in advance of the poster session. Posters should then be left up for the duration of the meeting. Materials for hanging posters will be near the poster board stands. Feel free to turn the posterboard stands to portrait conformation or to hang your posters from them if you need more room.

Oral presentations:

Please see the abstract book for the time and location of your talk. Presentations are either 15 or 30 minutes in length including question time. Session chairs will use timers to keep speakers on schedule.

Each room with have an audio microphone and a laser pointer. See your session chair for assistance with any AV equipment 15 minutes before the session.

Please load your presentation onto the embedded PC in the correct room and place your file in the appropriate session folder well in advance of the session. The computers will be logged in each morning. If for some reason they are logged off the username is "lecture". There is no password. Be sure to have the "this computer" pull down menu selected below before hitting enter.

