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Genetics Society of AustralAsia
55th Annual Meeting
Adelaide, Australia
7 - 10 July 2008

GSA
Genetics Society of AustralAsia Inc.

Information and Abstracts

Contents

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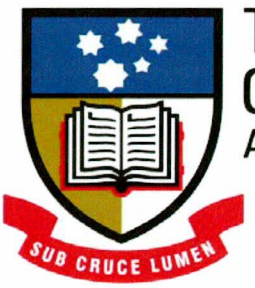

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HOW TO FIND THE VENUES

Map references to the map in your conference satchel

MIXER
Foyer, South Australian Museum Map Ref. M5

TALKS
NAPIER LECTURE THEATRES 1.02, LG28, LG29. Map Ref. K12
(Entrance to LG28, LG29 from Walter Young Garden Ref J11)

POSTERS AND COCKTAILS
Eclipse Room, Union House, Map Ref E5

CONFERENCE BANQUET
Pacific Cultures Gallery
South Australian Museum Map Ref. M5

ORGANISING COMMITTEE

CONVENOR: Jack da Silva, The University of Adelaide

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Invited Speakers

David Adelson, University of Adelaide
Eldon Ball, Australian National University
Matthew Bellgard, Murdoch University
Karen Burke da Silva, Flinders University
Brian Dalrymple, CSIRO
Sally Dunwoodie, Victor Chang Cardiac Research Institute
Jenny Graves, Australian National University
Richard Harvey, Victor Chang Cardiac Research Institute
Simon Ho, Australian National University
Margaret Katz, University of New England
Brett Lidbury, University of Canberra
Frederik Nijout, Duke University
David Penny, Massey University
Matt Phillips, Australian National University
Allen Rodrigo, University of Auckland
Ken Wolfe, Trinity College Dublin

Recommended nearby restaurants and cafes:

For a quick lunch:

Blue Lemon: 217 North Terrace (opposite Museum). Good choice of baguettes from $5.

Un Café Bar: 261 North Terrace (opposite Art Gallery). Good coffee, sandwiches and cakes.

Genki Roll Japanese, North Terrace downstairs in David Jones food court (opposite Art Gallery). Good take away sushi.

Aroma Café: cnr Frome Street and North Terrace. UniSA student café. Good coffee, sandwiches and salads.

Cibo Espresso: 218 Rundle St (cnr Frome St). Nice coffee & snacks. Wireless internet.

Art Gallery Restaurant: Located within the Art Gallery. Open 10-4.30pm. More upmarket café, very close.

Noodle Junction: Shop 12, 21 Pulteney St (between North Tce and Rundle Mall).

Popular spot for noodles.

Penang Hawkers Corner: Shop 9 Renaissance Arcade, 21 Pulteney St (enter arcade between North Tce and Rundle Mall). Good value Malaysian food, great for laksa.

For a more substantial lunch or dinner:

Café Michael 2 (8223 3519): 204 Rundle St. Open noon-2.30pm, from 5.30pm Thai food. Lunch specials from $10

Exeter Hotel (8223 2623): 246 Rundle St. Tasty pub food from about $10.

Amalfi Pizzeria (8223 1948): 29 Frome St, between North Tce and Rundle St. Open noon-2.30pm, from 5.30pm Mains $18-24. Best pizza in town. Can get busy.

Scoozi Caffe Bar (8232 4733): 272 Rundle St. Alternative italian café if you can't get into Amalfi's. Mains $17-20.

Jasmin Indian Restaurant (8223 7837): 31 Hindmarsh Sq corner of Pulteney and Grenfell St. Open from 5.30 Mains $20.50-$22.50. Tasty Indian dishes.

Eros Ouzeri (8223 4022): 277 Rundle St. Open noon-3, from 5.30 Mains $25-30. Traditional greek with modern fusion.

Cos Restaurant (8231 7611).: 18 Leigh St. Noon-3, 6-9 Mains $17-35. Modern Australian, Seafood and Steak.

Computer access at the conference

You will be informed of various forms of access to the internet during the conference. Call Internode on 13 NODE (13 66 33) if you need help with the wireless connection. Internet access is also available through computers in the Molecular Life Sciences Building, Room G.40 (Map Ref. C 11)

RNA-processing cascade. Richard Robinson.

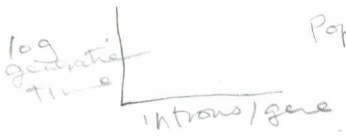
RNA Catalysis v slow. $1 min^{-1}$ Protein $10^{+} sec^{-1}$

euks. spliceosome. 6 RNAs 200? proteins.

SCUDS. Rare genomic changes

Inf for aa order not useful for deep phylogenies,

FRED had at least 4 introns/gene.



Spliceosome looks ancestral.

SOLEXA sequencer

FRED had a full RNA infrastructure? → primitive RNA system?

aa seq 5-700m yrs.

Ken Wolfe. Dublin

GENETICS SOCIETY OF AUSTRALASIA
ADELAIDE 2008
PROGRAM

Monday 7 July 2008
19.00 - 21.00 Mixer Foyer, South Australian Museum

Tuesday 8 July 2008

8.45 Opening, Napier Building, Lecture Theatre 1.02

Professor James McWha Vice Chancellor and President, The University of Adelaide

9.00 Plenary Lecture, Napier Building, Lecture Theatre 1.02

Chairperson: David Adelson

Frederik Nijout The Developmental Regulation and Evolution of Body size: Studies with *Manduca sexta* *Department of Biology, Duke University, USA* **Abstract 1**

10.00 – 10.30 Coffee break Lower Napier Building, Foyer

Symposium 1

Evolutionary Genetics,
Lower Napier Building LG29

Chairperson: Jeremy Timmis

10.30 David Penny, Lesley Collins; Sylvia Chen. *The ancestral eukaryote inferred from genomes* **Abstract 2**

11.00 Ken Wolfe. *Gene loss from a chloroplast mutation hotspot* **Abstract 3**

11.30 Anna E. Sheppard and Jeremy N. Timmis. *Plastid DNA instability in the nuclear genome* **Abstract 4**

11.45 Andrew H. Lloyd and Jeremy N. Timmis. *Functional gene transfer from the chloroplast to the nucleus in tobacco* **Abstract 5**

12.00 Deborah Shearman, Jennifer Morrow, Stuart Gilchrist, Kathie Raphael and Marianne Frommer. *Mating isolation in three species of tephritid fruit fly* **Abstract 6**

Concurrent Session 1:

Developmental Genetics and Cytogenetics
Lower Napier Building LG28

Chairperson: Stephen Donnellan

10.30 Janine E. Deakin, Edda Koina, Margaret L. Delbridge, Paul Waters, Amir Mohammadi, Jennifer A. Marshall Graves. *Efficient generation of a cytogenetic map of the tammar wallaby genome* **Abstract 7**

10.45 Ubon Tangkawanit, Chaliow Kuvangkadilok , Visut Baimai and Peter H Adler. *Cytogenetics of the Simulium tuberosum group (Diptera: Simuliidae) in Thailand* **Abstract 8**

11.00 Tamar Sztal, Henry Chung, Philip Batterham, Phillip J. Daborn. *The role of Cytochrome P0 genes in hormone synthesis and reproduction* **Abstract 9**

11.15 Warwick Grant, Susan Stasiuk and Matthew Crook. *Ecological genetics of Parastrongyloides trichisuri and the evolution of parasitism* **Abstract 10**

11.30 Balasubramanian Sureshkumar, Marco Todesco, Sridevi Sureshkumar, Janne Lempe and Detlef Weigel. *From QTL to gene: Exploiting natural variation in Arabidopsis thaliana to understand genetic basis of complex traits* **Abstract 11**

12.30 – 14.00 LUNCH (see page 7 for recommendations)

GSAA Committee Meeting during the lunch break (bring lunch to Lower Napier Building LG28)

Tuesday 8 July 2008

Symposium 2:

The Genetics of Development
Lower Napier Building LG29

Chairperson: Robert Richards

14.00 Owen WJ Prall, Mark J Solloway, Milena Furtado, Christine Biben, Daniel Schaft, Mary K Menon, Richard P Harvey. *Overlapping roles for Bmp/Smad signalling in heart development* **Abstract 12**

14.30 Sally L. Dunwoodie. *Notch Signalling, somitogenesis and abnormal vertebral segmentation* **Abstract 13**

15.00 Ben Tucker. *Fragile X Syndrome: genetic modelling and therapeutic leads in zebrafish* **Abstract 14**

15.15 Martin Lewis, Thomas Klaric, Colleen Bindloss, Muray Whitelaw, Michael Lardelli and Simon Koblar. *The Developmental Role of NPAS* **Abstract 15**

15.30 Stephen Gregory, Saman Ebrahimi, Joanne Milverton and Robert Saint. *Molecular mechanisms in cell division* **Abstract 16**

Concurrent Session 2:

Population and Evolutionary Genetics
Lower Napier Building LG28

Chairperson: Miguel de Barros Lopes

14.00 Hilary C. Miller, Jennifer A. Moore, Nicola J. Nelson, Charles H. Daugherty. *Do MHC genes influence mate choice in reptiles? A case study on tuatara (Sphenodon punctatus)* **Abstract 17**

14.15 Yuanyuan Cheng, Matthew Wakefield, Hannah Siddle, Penny C. Coggill, Cathy Herbert, Stephan Beck, Katherine Belov and Mark D. B. Eldridge. *Development of MHC-linked microsatellite markers in the tammar wallaby (Macropus eugenii)* **Abstract 18**

14.30 Jackie T. Chan. *Genetic estimates of dispersal and the implications for conservation management of grey-headed flying fox* **Abstract 19**

14.45 Sarah C.E. Bray, Jeremy Austin, Beth Shapiro, Jacobo Weinstock, Ian Barnes, Leopoldo Soibelzon, Alan Cooper. *Ancient DNA analysis of the extinct Tremarctine bears* **Abstract 20**

15.00 Gillian Gibb, Julia Goldberg, Steve Trewick, Ralph Powlesland, David Penny. *The Multiscale Nature of Phylogeny: A case study using the New Zealand Pigeon (Kereru)* **Abstract 21**

15.15 JY Yao, XC Jia, YC Crozier, YZ Chen, RH Crozier, JM Cook. *Self-pollination in an Australian fig species* **Abstract 22**

15.30 Beth E. Schlotfeldt, Terry Bertozzi, Steve Donnellan and Sonia Kleindorfer. *Adaptive divergence and gene flow in island and mainland populations of the Superb Fairy-wren (Malurus cyaneus) in South Australia* **Abstract 23**

15.45 Kym Ottewell, Andrew Lowe, Doug Bickerton and Phil Ainsley. *The genetic consequences of rarity: Case studies of South Australian endangered plant species and their common congeners* **Abstract 24**

16.00 Coffee break, Lower Napier Building, Foyer

16.05 Genetics Society of AustralAsia Annual General Meeting, Lower Napier Building LG28,
All members are encouraged to attend

17.00-19.00 POSTERS AND COCKTAILS Eclipse Room, Students' Union Building

Wednesday 9 July 2008

9.00 Plenary Lecture, Lower Napier Building, Lecture Theatre 1.02

Chairperson: Karen Burke da Silva

Patricia J. Pukkila, Martha S. Arnold, David H. Kiel.
Catalyzing Changes in Undergraduate Science Education **Abstract 26**

10.00 – 10.30 Coffee break, Lower Napier Building, Foyer

Symposium 3

Teaching Genetics and Evolution
Lower Napier Building LG29

Chairperson: David Catcheside

10.30 Brett A. Lidbury and Felicia Z. Zhang. *Language-Centred Pedagogy and the Impact on Learning in Undergraduate Genetics and Molecular Biology Units* **Abstract 27**

11.00 Karen Burke da Silva. *Teaching Evolution as core in First Year Biology* **Abstract 28**

11.30 John Sved. *The Hands On Genetics and other genetics and evolution computer teaching programs* **Abstract 29**

Concurrent Symposium 3

Molecular Systematics and Biogeography
Lower Napier Building LG28

Chairperson: Alan Cooper

10.30 Mark J. Sistrom. *Systematics and evolutionary history of the most successful Australian geckos – Gehyra* **Abstract 30**

10.45 Nic Rawlence, Trevor Worthy, Jeremy Austin, Alan Cooper. *Phylogeography of the extinct New Zealand moa Pachyornis: ancient DNA versus morphology* **Abstract 31**

11.00 Luke Price, Donnellan, S. and Mahony, M. *Patterns of diversification in northern Australian frogs* **Abstract 32**

11.15 Georgina M. Cooke, Ning L. Chao, Luciano B. Beheregaray. Water 'Colour' and Fish evolution: *A Comparative Phylogeographic Study of Amazonian Fishes across an Ecological Gradient* **Abstract 33**

11.30 Philippa Griffin. *Identifying distinct lineages among sympatric Australian alpine Poa species to predict evolutionary responses to climate change* **Abstract 34**

11.45 Jaro Guzinski. *Population genetics analysis reveals substructuring within a population of an Australian reptile tick Bothriocroton hydrosauri* **Abstract 35**

12.00 Robert A. B. Mason, Karen-Ann Gray, Rebecca N. Johnson, Catherine Price, Walter E. Boles. *Conservation genetics of the Bush Stone-curlew (Burhinus grallarius)* **Abstract 36**

12.15 Leo Joseph, Gaynor Dolman, Stephen Donnellan, Kathleen Saint, Mathew Berg and Andrew Bennett. *Where and When Does A Ring Start and End? Testing the Ring Species Hypothesis in a Species Complex of Australian Parrots* **Abstract 37**

12.30 David Alquezar, Robert D. Cooper, and Nigel W. Beebe. *Genomic analysis of the multicopied rDNA ITS in the mosquito Anopheles longirostris Burg (Diptera: Culicidae) and parallel assessment of the mtDNA cytochrome oxidase reveals eight cryptic species in Papua New Guinea* **Abstract 38**

12.45 – 14.00 LUNCH

Wednesday 9 July 2008

Symposium 4

Rates of Evolution
Lower Napier Building LG29

Chairperson: Jack da Silva

14.00 Allen Rodrigo and Peter Tsai. *Artifactual causes of substitution rate discrepancies* **Abstract 39**

14.30 Matthew J. Phillips. *Branch-length estimation bias misleads molecular dating* **Abstract 40**

15.00 Simon Y. W. Ho and Phillip Endicott. *Recalibrating the time-scale of human evolution using mitochondrial DNA* **Abstract 41**

15.30 Alan Cooper, Jack Da Silva. *The evolutionary rates curve: How accurate are molecular date estimates?* **Abstract 42**

Concurrent Session 4

Comparative genomics
Lower Napier Building LG28

Chairperson: Tasman Daish

14.00 Renfu Shao and Stephen C. Barker. *The head louse, Pediculus capitis, has an extraordinary mitochondrial genome* **Abstract 43**

14.15 Tatiana Vassilieva, Nagesh Chakka, Jill E. Gready. *Evolution of expression and function of the prion-protein family genes, PRNP and PRND* **Abstract 44**

14.30 Vidushi S. Patel, Steven J. B. Cooper, Janine E. Deakin, Bob Fulton, Tina Graves, Wesley C. Warren, Richard K. Wilson, Jennifer A. M. Graves. *A new model for the evolution of the globin gene clusters in amniotes* **Abstract 45**

14.45 Hardip Patel, Frickey, T., Waters, P., Delbridge, M., and Graves, J.A.M. *Comparative analysis of protein coding genes: Rearrangements and gene gain or loss* **Abstract 46**

15.00 Hannah S. Bender, Elizabeth P. Murchison, Margaret A. Strong, Daniel A. McMillan, Tariq Ezaz, Carol W. Greider, Anne-Maree Pearse, Gregory J. Hannon and Jennifer A. Marshall Graves. *Telomere length polymorphism in wild Tasmanian devils* **Abstract 47**

15.15 Kioumars Ghamkhar, Megan Ryan, Phil Nichols, Richard Snowball, Rudi Appels. *Genetics and ecogeographical studies in subclover (Trifolium subterraneum)* **Abstract 48**

15.30 Katherine Belov, Hannah Siddle, Jolanta Marzec, Claire Sanderson. *The role of the Major Histocompatibility Complex in Devil Facial Tumour Disease* **Abstract 49**

15.45 Emily Remnant, Phil Batterham and Phillip Daborn. *The expression, evolution and role of ligand-gated chloride channels in insecticide resistance* **Abstract 50**

16.00 – 17.00 Coffee break, Lower Napier Building, Foyer

17.00 The Sir Ronald Fisher Lecture, Napier Building, Lecture Theatre 1.02

Chairperson: Bob Hill, Executive Dean, Faculty of Sciences, The University of Adelaide

Ken Wolfe. *The impact of ancient polyploidisations on eukaryotic genome evolution* **Abstract 51**

Refreshments afterwards in the Foyer, Lower Napier Building

19.00 – 24.00 Conference Banquet Pacific Cultures Gallery South Australian Museum

non-melastran genes? ancient
coral specific genes.

Thursday 10 July 2008

9.00 Plenary Lecture, Napier Building, Lecture Theatre 1.02

Chairperson: Frank Grützner

Jennifer A. Marshall Graves. *Weird animal genomes and sex* **Abstract 52**

10.00 – 10.30 Coffee break, Lower Napier Building, Foyer

Symposium 5

Comparative Genomics
Lower Napier Building LG29

Chairperson: Stephen Gregory

10.30 Eldon E. Ball, David J. Miller. *Lower metazoan genomes-a window on genome evolution* **Abstract 53**

11.00 Matthew Bellgard. *An integrated approach to undertaking diverse studies in comparative genomics* **Abstract 54**

11.30 Frank Grützner and Enkhjargal Tsend-Ayush. *Large-insert genomic clones – a new tool to study genome organisation in sperm* **Abstract 55**

11.45 Janine E. Deakin, Timothy A. Hore, Edda Koina, Jennifer A. Marshall Graves. *The status of dosage compensation in the multiple X chromosomes of the platypus* **Abstract 56**

12.00 Camilla M. Whittington, Anthony T. Papenfuss, Katherine Belov. *From immunome to venome: Convergent evolution of platypus and reptile venom genes* **Abstract 57**

12.15 Henry Ye and Elizabeth A. McGraw. *The evolution of resistance to bacterial infection in Drosophila melanogaster* **Abstract 58**

Concurrent Session 5

Molecular Genetics
Lower Napier Building LG28

Chairperson: Louise O'Keefe

10.30 Michael Lardelli. *Truncated Presenilin peptides with potent dominant negative activity* **Abstract 59**

10.45 Kamlangdee, N, Lockington, RA, and Joan Kelly. *Targets of the CreB regulatory deubiquitinating enzyme in A. nidulans* **Abstract 60**

11.00 Tom Harrop, Philip Batterham and Phillip J. Daborn. *Assessment of Cytochrome P0 function in Drosophila melanogaster by RNA interference of the P0 redox partners CPR and Dare* **Abstract 61**

11.15 Jeremy R. Shearman and Alan N. Wilton. *Using the canine SNP array to identify disease genes* **Abstract 62**

11.30 Rugang Tian, Wayne Pitchford, Cynthia D.K.Bottema. *Molecular genetics of beef fat colour* **Abstract 63**

11.45 Bree Buszard, Tony Tiganis, and Coral Warr. *The Drosophila Protein Tyrosine Phosphatase dPTP61F regulates insulin signalling and growth* **Abstract 64**

12.00 Tasman Daish, Frank Grützner and Enkhjargal Tsend-Ayush. *Meiotic sex chromosome inactivation in monotremes* **Abstract 64a**

12.30 – 14.00 LUNCH

Thursday 10 July 2008

Symposium 6

Bioinformatics,
Lower Napier Building LG29

Chairperson: Martin Lewis

14.00 Brian. P. Dalrymple. *Identification and annotation of repeats in the bovine genome* **Abstract 65**

14.30 David L. Adelson, Joy M Raison, Robert C Edgar. *Building the sheep genome using comparative genomics and new generation sequencing technologies* **Abstract 66**

15.00 Gavin Huttley and Von Bing Yap. *Context and Codons: standard models are biased by sequence composition* **Abstract 67**

15.15 Lisa M. J. Bardsley, Wayn Wong, Charles Robin. *Pseudogenes in Drosophila* **Abstract 68**

15.30 William B Sherwin, Jurgis Sapijanskas & Dhriti Pandya. *Linkage disequilibrium in natural populations: information statistics provide a powerful test* **Abstract 69**

16.00 -16.30 Coffee break

16.30 M. J. D. White Address, Napier Building, Lecture Theatre 1.02

Chairperson: Jeremy Timmis

Margaret E. Katz. *Food, sex, drugs and death in fungi: identification of genes involved in nutrient sensing and programmed cell death* **Abstract 70**

17.30 – 18.00 Awards and Closing, Napier Building, Lecture Theatre 1.02

Fred Nijhout

Ken Wolfe · chloroplast = cyanobat 155kb circ

Rpl 32 → Cu Zn SOD ⟋ evolved alt. splicing

has translocation signal for chloroplast

ycf 4 lost in pea, +is a pseudogene, conserved in chl

sweet pea - intact, fnc i.e. photosynt I

Lathyrus

Tobacco vs cyanobact 47% aa identity

lathyrus vs taliyrus 31% aa

ABSTRACTS OF TALKS

1. The Developmental Regulation and Evolution of Body size: Studies with *Manduca sexta*

Frederik Nijout,

Department of Biology, Duke University, USA

Body size is one of the most characteristic features of species. In addition, evolution of body size is one of the most widespread trends in evolution, presumably due to its association with fitness. We have studied the developmental-physiological mechanisms that control body size in insects as diverse as Hemiptera, Coleoptera and Lepidoptera. The developmental mechanisms and internal cues that control body size are diverse and some apprear to be adapted to the life style of the insect.

The full sequence of events and checkpoints in the regulation of size are best understood in the tobacco hornworm Manduca sexta (Lepidoptera: Sphingidae), where we have been able to develop a fully predictive mathematical model for body size regulation. Simultaneous selection on body size and development time produces interesting evolutionary trajectories that can, in part, be explained by the developmental model.

Professor Nijhout has been at Duke University since 1977, where from 1991 to 1995 he served as chairman of the Zoology Department. His research has focused on the control of postembryonic development in insects, in particular the control of metamorphosis and of polyphenism, the development of alternative phenotypes from a single genotype. He has been particularly interested in the developmental and genetic mechanisms that control body size and the proportional sizes of body parts. He has also used theoretical and mathematical modeling to examine how non-linear processes in development, coupled with gene-environment interactions, affect our understanding of genotype-phenotype relationships.

2. The ancestral eukaryote inferred from genomes

David Penny, Lesley Collins, Sylvia Chen

Allan Wilson Center for Molecular Ecology and Evolution

Massey University, Palmerston North, New Zealand

With the increasing number of eukaryote genomes, and particularly from a wider selection of eukaryotes, it is becoming easier to infer the genome content of the last common ancestor of eukaryotes. One of the first established was the exon/introns structure of that genome, but now other properties are being inferred. These include the protein and small RNA composition of the spliceosome, one of the most complex structures in eukaryotes with 5 snRNAs and around 200 proteins in humans. In general, the composition of small RNAs appears to be quite extensive, including snoRNAs., and there is now some evidence for RNAi to be effectively universal in eukaryotes. The significance of the widespread occurrence of small RNAs will be discussed briefly.

3. Gene loss from a chloroplast mutation hotspot.

Ken Wolfe

Smurfit Institute of Genetics, Trinity College Dublin, Ireland.

The chloroplast genomes of flowering plants usually have a highly conserved structure with the same 79 protein-coding genes being present, and in the same order, in almost all the ~75 sequenced angiosperm cpDNAs. A few plant families, however, show substantial divergence from this pattern. One such family is the legumes (Fabaceae). By sequencing the chloroplast genomes of pea (*Pisum sativum*, in collaboration with John Gray at Cambridge) and grasspea (*Lathyrus sativus*) we find that both of these genomes are extensively rearranged (8 and 6 inversions, respectively, relative to the angiosperm consensus organization). Both genomes also lack several genes that are normally present in cpDNA: *infA*, *rps16*, *rpl22*, and *rpl23* are absent in both species, *ycf4* is a pseudogene in pea, and *psaI* is absent in grasspea. The latter two are the first known losses of photosynthesis-related genes from cpDNA of photosynthetic angiosperms. The *ycf4*, *psaI* and *rps16* loci are located close together in legumes. Analysis of a 5 kb region that includes these loci and one other gene (*accD*) shows that it has a mutation rate at least 10 times higher than in the rest of the genome. This hotspot has caused very rapid evolution of the genes that remain in it: there is more sequence divergence between the Ycf4 proteins of two species in the genus *Lathyrus* than between tobacco and a cyanobacterium. We hypothesise that the hotspot also caused *psaI*, *rps16* and *ycf4* (in pea) to be lost from the genome. We were unable to find transferred copies of *ycf4*, *psaI* or *rps16* in the nuclear genome, and hypothesise that they have been lost completely.

Anna Shepherd Plastid instability in mal gene

Freq of gene transfer chl → nucl.

nea w mal parasite → chl.

backcross to wt ♀ high trans. freq. 1/16,000

Kr 2.1 @ 75% kan resist progeny - stable.

Kr 2.5 instability 4 inds.

1 ind Kr 2.9 - all redn mutic

Connor McNulla.

Wolbachi adaptation to its host

after long term serial passage in

mosquito cell lines

4. Plastid DNA instability in the nuclear genome

Anna E. Sheppard and Jeremy N. Timmis

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

Gene transfer from the plastid (chloroplast) and mitochondrial genomes to the nucleus has been an important driving force in eukaryotic evolution. Non-functional DNA transfer is far more frequent and the frequency of such transfers from the plastid to the nucleus has been determined experimentally in tobacco using transplastomic lines containing in their plastid genome a kanamycin resistance gene (*neo*) readymade for nuclear expression. Contrary to expectations, non-Mendelian segregation of the kanamycin resistance phenotype is seen in progeny of some of these lines containing *neo* in their nuclear genome. Nine of these lines have now been analysed in detail and the instability has been shown to be due to the loss of *neo*. Four lines showed instability with variation between progeny derived from different areas of the same plant, suggesting a loss of *neo* during somatic cell division. One line showed a consistent reduction in the proportion of kanamycin resistant progeny, suggesting a loss of *neo* during meiosis, and the remaining four lines appeared to be stable. The work described here provides evidence that insertion and removal of plastid DNA in the nuclear genome are in dynamic equilibrium. These processes may be important in driving the evolution of new nuclear genes.

5. Functional gene transfer from the chloroplast to the nucleus in tobacco

Andrew H. Lloyd and Jeremy N. Timmis

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

DNA transfer from the organelles (mitochondria and plastids) to the nucleus has been a major driving force of eukaryotic nuclear genome evolution. The vast majority of DNA transfer to the nucleus is non-functional. However, in a small subset of transfer events the gene is activated and functional gene transfer established. While significant steps have been made in elucidating the frequency of organelle to nucleus DNA transfer, less is understood about gene activation upon arrival in this new environment. The work presented here has enabled experimental observation of these evolutionary events and molecular analysis of the steps leading to activation. We had previously generated a number of independent tobacco lines in which a segment of the chloroplast genome had been transferred to the nucleus. Twelve of these lines were screened for nuclear activation of a chloroplast transgene (*aadA*). From the ~1.5 billion cells screened 3 plants were generated that showed spectinomycin resistance due to activation of *aadA* through acquisition of a nuclear promoter. In at least one case this resistance was mitotically unstable. The results presented suggest that functional gene transfer involves a dynamic interplay between transfer, rearrangement, function and loss of nuclear organelle DNA sequences.

6. Mating isolation in three species of tephritid fruit fly

Deborah Shearman, Jennifer Morrow, Stuart Gilchrist, Kathie Raphael and Marianne Frommer

School of Biological Sciences, A12, University of Sydney, NSW 2006, Australia.

Australian fruit flies (family Tephritidae) include our most serious horticultural pests and also provide intriguing problems of speciation. The sympatric pest species *Bactrocera tryoni* and *Bactrocera neohumeralis* show a clear mating isolation mechanism based on time of mating, but are so closely related that no fixed sequence difference has yet been found in any gene or microsatellite allele isolated to date. Using hybrid lines generated from forced mating between the two species, we have shown that the mating isolation mechanism involves differential expression of genes with diurnal cycling patterns. A third species, *Bactrocera jarvisi*, has a geographic distribution and host fruit preference that overlap with *B. tryoni* and *B. neohumeralis*. *B. jarvisi* is strongly differentiated from *B. tryoni* and *B. neohumeralis*, in mitochondrial genes, nuclear genes and microsatellite sequences. Nonetheless, *B. jarvisi* mates readily with *B. tryoni*, to produce viable and fertile hybrids. We have yet to establish a molecular basis for mating isolation between *B. tryoni* and *B. jarvisi* in the wild, but the lack of mating isolation in the laboratory has allowed us to use *B. jarvisi* genomic sequences to generate marked strains of *B. tryoni* for Sterile Insect Release and for sexing of embryos and larvae.

7. Efficient generation of a cytogenetic map of the tammar wallaby genome

Janine E. Deakin, Edda Koina, Margaret L. Delbridge, Paul Waters, Amir Mohammadi, Jennifer A. Marshall Graves

ARC Centre for Kangaroo Genomics, Research School of Biological Sciences, The Australian National University, Canberra ACT 0200, Australia

Marsupials have proven to be extremely valuable inclusions in comparative genomic studies. To this end, two distantly related model marsupials have been sequenced: the South American opossum (*Monodelphis domestica*) and the tammar wallaby (*Macropus eugenii*), which last shared a common ancestor ~70 MYA. The opossum genome assembly (x6 coverage) has been assigned to chromosomes, but with the wallaby genome having been sequenced to only a depth of two-fold, the assignment of sequence to chromosomes by mapping only ultracontigs from this assembly would leave a large amount of sequence unanchored. To overcome this problem, we devised an efficient strategy to generate a cytogenetic map of the wallaby genome. This approach is based on chromosome homology revealed by cross-species chromosome painting and using the opossum assembly to predict which genes lie on each wallaby chromosome. By aligning the opossum genome assembly to the human assembly, we have identified large blocks of shared synteny. To locate and orient these blocks, we chose conserved genes at the end and within each block, then used sequence from the wallaby genome sequencing project to design specific overgo probes to isolate homologous wallaby BACs for cytogenetic mapping. This has proved to be a very rapid and efficient approach.

8. Cytogenetics of the *Simulium tuberosum* group (Diptera: Simuliidae) in Thailand.

Ubon Tangkawanit, Chaliow Kuvangkadilok, Visut Baimai and Peter H Adler

Department of Biology, Faculty of Science, Mahidol University, Bangkok, 10400, Thailand.

The polytene chromosomes of 3,347 larvae of the *Simulium tuberosum* group collected from 59 locations in northern, northeastern, central and southern Thailand were analyzed. Band by band comparisons, relative to the established standard chromosome map for the subgenus *Simulium*, revealed 17 cytogenetically distinct taxa, based mainly on fixed, sex-linked and floating inversions in the long arm of chromosome III. Six of these taxa correspond to morphologically described species, i.e., *S. doipuiense*, *S. rufibasis*, *S. setsukoae*, *S. tani*, *S. yuphae* and *S. weji*; 2 unknown species (unknown sp.1 and unknown sp.2) were discovered. Two cytoforms (cytoform A and B) were found in *S. doipuiense* based on different sex-linked inversions, whereas 9 cytoforms (cytoforms A-I) were present in *S. tani*. *S. tani* cytoforms A and E differed from the standard cytoform (cytoform B) by fixed inversions. Cytoforms C, D, F, G and H were distinguished by sex-linked inversions, X_2Y_1, X_0Y_1, X_2Y_0, X_1Y_2 and X_3Y_0, respectively. Cytoform I was characterized by floating inversions. Shared unique chromosomal features, relative to the subgeneric standard chromosome map allowed evolutionary relationships among the cytotaxa to be inferred. Ecologically, the distributions of larvae of each taxon may be influenced by some macro- and microhabitat factors.

Tangkawanit U, Kuvangkadilok C, Baimai V and Adler PH. Cytosystematics of the *Simulium tuberosum* (Diptera: Simuliidae) in Thailand. *Zool J Linn Soc.* ., (In press)

9. The role of Cytochrome P450 genes in hormone synthesis and reproduction

Tamar Sztal, Henry Chung, Philip Batterham, Phillip J. Daborn

Centre For Environmental Stress and Adaptation Research (CESAR), Department of Genetics, Bio21 Molecular and Biotechnology Institute, The University of Melbourne, Victoria, 3010, Australia

Cytochrome P450s are a large multigene family with members found in virtually all organisms. While it is postulated that the primary role of many P450s is detoxification, there are many involved in essential developmental processes, such a hormone synthesis. In an effort to identify P450s involved in important developmental functions in insects, we have characterized the expression patterns of all 85 *Drosophila melanogaster* P450s in embryos and third instar larvae. We show that many P450s are expressed in specific organs such as the brain, gonads or the oenocytes and that this expression is conserved throughout development. We have identified a single gene, *Cyp6g2*, which is expressed in the corpora allata, the site of juvenile hormone synthesis. RT-PCR indicates that *Cyp6g2* expression is correlated with times of juvenile hormone synthesis and is influenced by mating. RNAi analysis of *Cyp6g2* resulted in a lethality phenotype suggesting a more specific role in insect hormone production.

Richard Harvey. Role of Bmps.

- Nkx2-5 is central. feedforward + Thresholds.
- Bistable systems. - feedback loops.
- Nodal / lefty (1 & 2)

10. Ecological genetics of *Parastrongyloides trichisuri* and the evolution of parasitism

Warwick Grant[1,2], Susan Stasiuk[1] and Matthew Crook[1,2]

[1]Genetics Department, Latrobe University, Bundoora, 3086, Australia

[2]The Hopkirk Research Institute, AgResearch, Palmerston North, New Zealand

Parasitism as a life history strategy has arisen several times in the Phylum Nematoda, prompting the hypothesis that some feature of the basic nematode life cycle facilitates this evolutionary step. The dauer larva, a specialized facultative survival stage in the life cycle of free living nematodes, is proposed to play a central role in this process. We have studied the biology and genetics of a dauer-like switch in the parasitic nematode *Parastrongyloides trichosuri*. Nematodes of the genus *Parastrongyloides* are unique in that they are capable of free-living as well as parasitic life cycles, and can be considered as facultative parasites (1). The transition from one life cycle to the other may have been under selection during the evolution of parasitism in this group of organisms. We present data that shows that the insulin signaling pathway is most likely a key regulator of this transition, and evidence that there is genetic variation in the population for the signal response of this pathway to environmental conditions.

(1) Grant, W. N *et al.*,(2006). *Parastrongyloides trichosuri*, a nematode parasite of mammals that is uniquely suited to genetic analysis. International Journal for Parasitology, 36, 453-466

11. From QTL to gene: Exploiting natural variation in *Arabidopsis thaliana* to understand genetic basis of complex traits.

Sureshkumar Balasubramanian[#$], Marco Todesco[$], Sridevi Sureshkumar[#$], Janne Lempe[$] and Detlef Weigel[$Ψ].

[#] School of Integrative Biology, The University of Queensland, Brisbane, Australia

[$] Max-Planck Institute for Developmental Biology, Tuebingen, Germany

[Ψ] Salk Institute for Biological Studies, USA.

Deciphering the genetic and mechanistic basis of complex traits remains a formidable challenge. We are exploiting natural variation in *Arabidopsis thaliana* to unravel the genes that contribute to phenotypic variation in natural populations. The availability of a large number of strains that display extensive phenotypic variation, almost unlimited availability of markers and the extensive genetic and genomic tools make natural variation in *Arabidopsis thaliana* an attractive system to address the genetic basis of complex traits and their interactions with environment at a mechanistic level. I will present a brief overview of the use of natural variation and provide examples from our work describing the natural variation in phenotypes, identification of genes that underlie this variation and their evolutionary implications.

12. Overlapping roles for Bmp/Smad signalling in heart development

Owen WJ Prall, Mark J Solloway, Milena Furtado, Christine Biben, Daniel Schaft, Mary K Menon, Richard P Harvey

Victor Chang Cardiac Research Institute, Sydney 2010, Australia

Congenital heart disease (CHD) is the most common cause of non-infectious death in the first year of life, although its genetic causation is poorly understood. Our laboratory studies the cardiac developmental hierarchies and their involvement in CHD using the mouse model. Signalling through bone morphogenic proteins (Bmps) and effector transcription factors of the Smad family have diverse and overlapping roles in cardiogenesis. Bmps induce the myocardial lineage, and Bmp-Smads directly activate cardiac transcription factors such as the homeodomain factor Nkx2-5. Bmps are subsequently involved in valve and chamber formation. We have now shown that the earliest function for Nkx2-5 is as a transcriptional repressor, targeting *Bmp2* and other progenitor genes, and this pathway, when disrupted, is causative for CHD. In an integrated fashion, Bmp signalling through Smad1 is also responsible for setting a threshold for activation of the Nodal pathway that determines the left/right handedness of body organs including the heart.

13. Notch Signalling, somitogenesis and abnormal vertebral segmentation

Sally L. Dunwoodie

Developmental Biology Division, Victor Chang Cardiac Research Institute, Sydney, 2010 Australia

Somites are the precursors of the vertebral column. During embryogenesis they segment from the presomitic mesoderm (PSM) that is caudally located and newly generated from the tailbud. Somites form in synchrony on either side of the

embryonic midline in a reiterative manner. A molecular clock that operates in the PSM drives this process and controls the reiterative activity of the Notch signalling pathway. Superimposed on this reiterative activity is tight spatial regulation of Notch signalling in the PSM, which is achieved through the activity of modifiers of Notch signalling. Notch signalling is modified through the interplay of ligands that both *trans*-activate and *cis*-inhibit signalling and as a result of receptor gylocosylation by Fringe. Disruption of Notch signalling in the PSM impacts on somite formation causing abnormal vertebral segmentation (AVS). In mouse, Notch signalling components critical for somitogenesis have been identified. In humans, spondylocostal dysostosis (SCD), an abnormal vertebral segmentation syndrome is caused by mutation in Notch pathway genes. The mechanics of Notch signalling in the PSM will be discussed as will the role of Notch in SCD.

14. Fragile X Syndrome: genetic modelling and therapeutic leads in zebrafish

Ben Tucker

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

The fragile X mental retardation syndrome (FXMR) is caused by expansions of a CGG repeat in the *FMR1* gene. The *FMR1* related gene family is well described in human, mouse *Drosophila* and recently in zebrafish. Expression of the *FMR1*-related gene family in zebrafish has been found to be consistent with expression in mouse and human, implying that zebrafish should be an excellent model organism in which to study the vertebrate *FMR1*-related gene family. Knockout of the *FMR1* gene in mouse, drosophila and in FXMR results in distinct and well conserved abnormalities in dendritic and dendritic spine morphology. Behavioral abnormalities are found in the *FMR1* knockout mouse, related to defective long term depression through mGluR signaling. A subset of these behavioral abnormalities can be ameliorated by application of MPEP, an mGluR5 antagonist. This suggests that exaggerated mGluR signaling underlies symptoms of FXMR. We have demonstrated that similar dendritic abnormalities are found in zebrafish fmr1 morphants. We are now using the zebrafish to study whether the mGluR5 antagonists affect the dendritic structural abnormalities related to knockdown of *fmr1*. This will describe the level at which the mGluR5/ *FMR1* interaction is important in relation to synaptic plasticity, rather than at the behavioral level.

15. The Developmental Role of NPAS4

Martin Lewis, Thomas Klaric, Colleen Bindloss, Muray Whitelaw, Michael Lardelli and Simon Koblar

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

NPAS4 is a member of the basic-Helix-Loop-Helix / Per Arnt Sim transcription factors. This bHLH / PAS family includes the hypoxia-inducible-factors, single-minded 1 and 2, the dioxin receptor, and clock. The bHLH and PAS domains enable dimerisation and sensing of the environment. Once a heterodimer is formed a functional transcriptional regulator can bind nucleotide motifs specific to the regulatory protein. NPAS4 expression is restricted to brain with a very small level detected in testis. Further characterization finds NPAS4 expression is predominantly seen in the neurogenic tissues of brain.

NPAS4 is induced by trauma to the brain; examples of this include seizure (Flood et al. 2004) cerebral ischaemia and cortical spreading depression. Reduced expression of NPAS4 has been shown to occur when mice were reared in social isolation. We are investigating of the developmental role of NPAS4 in the model organism *Danio rerio.* Knock-down of NPAS4 by morpholino injection resulted in disruption of forebrain development. The specific changes in brain development supporting the hypothesis that NPAS4 is involved in neurogenesis will be presented.

Flood WD, Moyer RW, Tsykin A, Sutherland GR, Koblar SA. (2004) Nxf and Fbxo33: novel seizure-responsive genes in mice. *Eur J Neurosci.*, 20(7): 1819-26.

16. Molecular mechanisms in cell division

Stephen Gregory, Saman Ebrahimi, Joanne Milverton and Robert Saint

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

The mitotic microtubule array plays two primary roles in cell division. It acts as a scaffold for the congression and separation of chromosomes, and it specifies and maintains the contractile ring position. The current model for initiation of Drosophila and mammalian cytokinesis postulates that equatorial localization of the RhoGEF Pebble by a

microtubule-associated motor protein complex creates a band of activated RhoA, which subsequently recruits contractile ring components such as actin, myosin and Anillin. Equatorial microtubules are essential for continued constriction, but how they interact with the contractile apparatus is unknown.

We report the first direct molecular link between the microtubule spindle and the acto-myosin contractile ring. We find that the spindle-associated component, RacGAP50C, which specifies the site of cleavage, interacts directly with Anillin, an actin and myosin binding protein found in the contractile ring. Both proteins depend on this interaction for their localisation. In the absence of Anillin, the spindle-associated RacGAP loses its association with the equatorial cortex and cytokinesis fails. These results account for the long-observed dependence of cytokinesis on the continual presence of microtubules at the cortex.

17. Do MHC genes influence mate choice in reptiles? A case study on tuatara (*Sphenodon punctatus*).

Hilary C. Miller, Jennifer A. Moore, Nicola J. Nelson, Charles H. Daugherty

Allan Wilson Centre for Molecular Ecology and Evolution, School of Biological Sciences, Victoria University of Wellington, PO Box 600, Wellington, New Zealand

Major Histocompatibility Complex (MHC) genes are highly polymorphic components of the vertebrate immune system that play a key role in pathogen resistance. MHC genes may also function as odour-related cues for mate choice, thus ensuring optimal MHC diversity in offspring. MHC-associated mate choice has been demonstrated in some fish, bird and mammal species but there is little evidence for its occurrence in reptiles and it is not known whether this is a general vertebrate phenomenon. We investigated whether MHC-associated mate choice occurs in a wild population of tuatara (*Sphenodon punctatus*), a territorial and sexually dimorphic reptile. There was no significant association between male mating success, number of MHC sequences, microsatellite heterozygosity, or MHC lineage. In addition, no difference in the number of shared MHC alleles between mated and random pairs was observed. However, we did observe a trend towards individuals preferring mates with a functionally different genotype to their own, based on amino acid genotypic distance between pairs. The major determinant of mating success in tuatara was male body size, which was not related to MHC lineage or microsatellite heterozygosity. Our data suggest that in species such as tuatara, where mating patterns are primarily driven by male competitive ability, MHC-associated factors may play a lesser role.

18. Development of MHC-linked microsatellite markers in the tammar wallaby (*Macropus eugenii*)

Yuanyuan Cheng[1], Matthew Wakefield[2, 3], Hannah Siddle[1], Penny C. Coggill[4], Cathy Herbert[1], Stephan Beck[5], Katherine Belov[1*] and Mark D. B. Eldridge[6]

1. Faculty of Veterinary Science, The University of Sydney, NSW 2006, Australia.2. Bioinformatics, The Walter and Eliza Hall Institute, Parkville, VIC 3050, Australia.3. ARC Centre of Excellence for Kangaroo Genomics, Department of Zoology, The University of Melbourne, Parkville, VIC 3010, Australia.4. Wellcome Trust Sanger Institute, Wellcome Trust Genome Campus, Hinxton Hall, Hinxton, Cambridgeshire, CB10 1SA, UK.5. University College London, London WC1E 6BT, UK. 6. Australian Museum, 6 College St, Sydney, NSW 2010, Australia.

The Major Histocompatibility Complex (MHC) contains genes which play a significant role in immune response and mate choice, and are therefore of functional importance to molecular geneticists. The MHC of the tammar wallaby is highly unusual, with the Class I genes dispersed to ten locations on six chromosomes instead of clustering together in a single genomic region (Deakin *et al.*, 2007). In this study, we have developed ten microsatellite markers that are linked to MHC Class I genes which map to five different chromosomes in the tammar wallaby. All ten loci are highly polymorphic, with the expected heterozygosity ranging from 0.547 to 0.919. These microsatellites will assist in evaluating levels of MHC diversity in the tammar wallaby, understanding the impact of selection on genetic variation in tammar wallaby populations, and investigating the unique structure of the MHC of this marsupial. Six loci will also be useful in genetic and population studies in other macropod species.

Deakin JE, Siddle HV, Cross JGR, Belov K, Graves JAM (2007) Class I genes have split from the MHC in the tammar wallaby. *Cytogenetic and Genome Research*, 116: 205-211

19. Genetic estimates of dispersal and the implications for conservation management of grey-headed flying fox

Jackie T. Chan

School of Biological, Earth and Environmental Science, University of New South Wales, Sydney 2052 Australia.

The grey-headed flying fox (*Pteropus poliocephalus*) is currently listed as vulnerable and is protected in some areas. However, culling is still tolerated in some areas due to crop damage caused by the animal. Under these circumstances effective conservation management of large-range migratory species such as flying foxes requires considerable understanding of genetic interactions between populations. The current management strategies are incompatible unless there is very limited dispersal between populations. Estimates of dispersal can be obtained by measuring genetic variation amongst the populations and interactions between populations can be quantified. This study made genetic estimates of dispersal between grey-headed flying fox populations covering the speices range. The results suggested the grey-headed flying populations should be considered as a single panmictic population. Potential influences of selection, demographic history and introgression were also investigated, and results showed that these factors are unlikely to invalidate the inferred dispersal and population structure. The inferred dispersal can provide inputs to the demographic modeling of the species, to assist future conservation planning of the species. The model could possibly be used to make predictions about other flying foxes species.

20. Ancient DNA analysis of the extinct Tremarctine bears

Sarah C.E. Bray, Jeremy Austin, Beth Shapiro, Jacobo Weinstock, Ian Barnes, Leopoldo Soibelzon, Alan Cooper

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

The Tremarctinae are a New World subfamily of bears containing 4 known genera originating in the Pliocene (*Plionarctos*, *Arctodus*, *Arctotherium* and *Tremarctos*), of which three are now extinct. The only surviving member of the Tremarctine lineage is the South American *Tremarctos ornatus* (spectacled bear), an arboreal, primarily vegetarian, bear that is associated with high altitude Andean cloud forests.

In contrast, the fossil record suggests that at least two hyper-carnivorous members of the Tremarctine radiation, the North American extinct *Arctodus simus* (giant short-faced bear) and its South American relative *Arctotherium sp.* inhabited the New World until as recently as 21,000 – 10,000 years ago. However, the evolutionary and ecological relationships of these taxa remains unclear.

This study uses ancient DNA techniques to retrieve mitochondrial DNA sequences from >20 specimens of extinct Tremarctine bears across their known range. Preliminary results indicate a large genetic divergence between the extinct North American and South American genera, and a surprising lack of diversity within the North American giant short-faced bear. Additionally, and in marked contrast to ancient DNA analyses of North American Pleistocene brown bears (*Ursus arctos*) (Barnes *et al.*, 2002), the giant short-faced bears do not appear to exhibit strong phylogeographic structure.

Barnes I, Matheus P, Shapiro B, Jensen D, Cooper A. (2002) Dynamics of Pleistocene Population Extinctions in Beringian Brown Bears. *Science*. 295: 2267-2270

21. The Multiscale Nature of Phylogeny: A case study using the New Zealand Pigeon (Kereru)

Gillian Gibb, Julia Goldberg, Steve Trewick, Ralph Powlesland, David Penny

Allan Wilson Centre for Molecular Ecology and Evolution, Massey University, New Zealand

Evolutionary relationships are studied at a wide range of time scales, though little formal attention is paid to the multiscale nature of the process. In particular, the standard claim of Darwinian evolution that microevolutionary processes are sufficient to explain macroevolution requires consideration of time scales from processes in populations to the origin of major groups. Here we use DNA to integrate studies over the full range from dispersal of individual birds to the avian tree of life. We suggest such integration can be achieved relatively painlessly and without acrimony, though some changes in analytical approaches are required. As an example of an integrated study we use the New Zealand pigeon, Hemiphaga novaeseelandiae, and estimate its genetic diversity throughout its range, including the Chatham Islands and Norfolk Island (where it is now extinct). We use longer sequences to identify its nearest relatives within the South Pacific (including the imperial pigeons [Ducula] and fruit doves [Ptilinopus]), and its position within Columbidae (pigeons and doves) generally. Finally,

we use its complete mitochondrial genome, together with a sandgrouse (Pterocles namaqua), to study the position of pigeons within the Neoaves radiation. At the deeper levels of phylogeny we wish to reduce the noise in the data and enhance the signal, leading to clearer resolution of the basal nodes of avian phylogeny.

22. Self-pollination in an Australian fig species

JY Yao, XC Jia, YC Crozier, YZ Chen, RH Crozier, JM Cook

School of Marine and Tropical Biology, James Cook University, Townsville, Queensland, Australia 4811

Inbreeding is prevented in fig trees by synchronous production of flowers within a tree, but in some species there is asynchronous flowering raising the possibility of self-pollination through pollinator wasps entering syconia from their natal tree. We tested whether self-pollination actually occurs in such cases of asynchronicity. Microsatellite analysis using six highly-polymorphic loci showed that six seedlings of 112 from *Ficus rubiginosa* trees in asynchronous flowering periods were apparently inbred, but none of 108 from such trees during periods of strictly synchronous crops. Given the levels of polymorphism of these loci, these results are highly significant and show for the first time that periods of crop asynchrony for single trees do yield inbred progeny naturally. One apparently inbred progeny out of 100 was found in a sympatric and more strictly synchronous species, *F. racemosa*, but this is within random expectation given the levels of polymorphism for the eight loci used for this species. Given that experimental self-pollination was not found to reduce fitness in another fig species, and that fig seeds are typically deposited in clumps by frugivores, improved herd immunity from outbreeding is suggested as selective factor favouring outbreeding for future work. Estimates of the number of pollen parents per sibship were significantly higher for *F. rubiginosa* than for *F. racemosa*.

23. Adaptive divergence and gene flow in island and mainland populations of the Superb Fairy-wren (*Malurus cyaneus*) in South Australia.

Beth E. Schlotfeldt[1], Terry Bertozzi[2], Steve Donnellan[2] and Sonia Kleindorfer[1]

[1] *School of Biological Sciences, Flinders University, South Australia, Australia.*

[2] *Evolutionary Biology Unit, South Australian Museum, Adelaide, Australia*

Environmental gradients are thought to drive adaptive phenotypic changes between populations in varying environments (sometimes in the face of the homogenising effects of gene flow) and may therefore be a precursor to speciation. For example, changes in allele frequencies may follow shifts in behavioural and morphological characters (as a result of differential selection acting on genetically variable traits). The level of gene flow and strength of selection may determine the degree of adaptedness of organisms to their local environment. Habitat differences (e.g., vegetation structure, prey availability, local climatic conditions) and strength of selection may determine the relative influence of gene flow and adaptive divergence in sedentary birds such as the Superb Fairy-wren.

This project examines morphology, foraging ecology, and prey availability data for evidence of adaptive divergence and microsatellite variation for evidence of gene flow across Kangaroo Island and mainland Mount Lofty Ranges populations of Superb Fairy-wrens in South Australia.

24. The genetic consequences of rarity: Case studies of South Australian endangered plant species and their common congeners

Kym Ottewell[1], Andrew Lowe[12], Doug Bickerton[2] and Phil Ainsley[3]

[1] *The University of Adelaide, School of Earth and Environmental Sciences, North Terrace, Adelaide, 5005, South Australia, Australia.* [2] *Department of Environment and Heritage, GPO Box 1047, Adelaide, 5000, South Australia, Australia.* [3] *Botanic Gardens of Adelaide, Seed Conservation Centre, North Terrace, Adelaide, 5000, South Australia, Australia*

Comparisons of population genetic diversity between related rare and widespread species can provide valuable insights into the consequences of rarity and are critical for guiding conservation planning and restoration efforts. We are currently investigating the conservation genetics of ten rare and threatened South Australian plant species and their common congeners, encompassing species from eight genera and five families, to gain an understanding of the genetic consequences of small population size and population isolation. Here we present results for two critically endangered species, *Prostanthera eurybioides* (Monarto Mintbush) and *Acacia pinguifolia* (Fat-leaved Wattle), both of which have disjunct distributions and are known from small fragmented populations. We compare the levels of genetic

diversity and population differentiation of these rare species with their common and more widespread common congeners, *P. behrii* and *A. halliana,* to help identify potential management options to address issues of recruitment and population decline.

25. withdrawn

26. Catalyzing Changes in Undergraduate Science Education

Patricia J. Pukkila,[1,2] Martha S. Arnold,[2] David H. Kiel[3]

[1]Department of Biology, University of North Carolina at Chapel Hill, Chapel Hill, NC, [2]Office for Undergraduate Research, University of North Carolina at Chapel Hill, Chapel Hill, NC, [3]Institute for the Arts and Humanities, University of North Carolina at Chapel Hill, Chapel Hill NC

To make research a distinctive feature of the undergraduate experience at UNC-Chapel Hill, we have implemented a variety of programs designed to support faculty as they reflect on approaches to research, and incorporate inquiry-based methods and research experiences into their courses. Faculty are invited to participate in a multidisciplinary undergraduate seminar course ("Modes of Inquiry") in which they discuss their research interests, particularly how topics are narrowed and "points of entry" to particular investigations are defined. Faculty are also invited to utilize inquiry-based teaching methods, which focus on helping students to make a transition from "novice-like" approaches to learning (memorizing conclusions reached by others) to "expert-like" approaches (a procedural understanding of how current conclusions were reached). In addition, faculty are invited to modify existing courses to include student research components with the assistance of a collaborating graduate student (the "Graduate Research Consultant"). Taken together, these programs encourage individual innovations, require reflection and assessment of the results, and provide opportunities for faculty to influence other faculty. In our opinion, this deliberate framework that includes multiple levels of faculty involvement is essential to catalyze meaningful and lasting change, and we believe that our approach could be adopted successfully in a wide variety of educational settings. (Supported by the NSF.)

Professor Pukkila has been at the University of North Carolina at Chapel Hill since 1979. In 1999, she became the founding Director of the University's Office for Undergraduate Research. Her research has focused on meiotic chromosome behavior and fungal genomics. She has been particularly interested in recombination "hot spots" and their role in meiotic chromosome pairing, and she heads the Coprinus cinereus genome project. She has been recognized for her contributions to science education at the local, state and national levels. In 1996, Pukkila established the "Genetics Education" section of the journal Genetics, which has provided a scholarly showcase for innovations in teaching and learning genetics. And in 2001 she initiated biannual, multi-campus undergraduate research symposia for the North Carolina state legislature. These ongoing symposia enable students to convey the excitement and importance of their original work to their elected officials. She is a Fellow of the American Association for the Advancement of Science.

27. Language-Centred Pedagogy and the Impact on Learning in Undergraduate Genetics and Molecular Biology Units

Brett A. Lidbury and Felicia Z. Zhang.

Faculty of Science and Faculty of Communication and International Studies, The University of Canberra, ACT, 2601 Australia.

A barrier to undergraduate learning in *Genetics* and *Molecular Biology* is the language of these disciplines. In this study, learning interventions were designed to incorporate foreign language education techniques, via collaboration with an applied linguist and lecturer in Mandarin Chinese, in the teaching of these units. We report on the findings of semesters where the Genetics lecturer and Language lecturer worked in partnership on the delivery and design of foreign language interventions for *Genetics* and *Molecular Biology* students. Interventions included on-line genetic language exercises using *Hot Potatoes*™ software, emphasis on the structure of scientific papers, translation of difficult scientific words and concepts identified in genetics papers, and lecture/tutorial warm-up language exercises. Student performance on assessment tasks versus prior "non-language" student cohorts and survey results of student experience were analysed. Language intervention was found to enhance academic performance for Distinction (DI) students, with no statistically significant effect found for Pass/Credit students. Surveys indicated that on-line assessment, revising concepts in tutorials through small group work and opportunities for students to provide different points of view were the most popular language supporting activities with students. We also report on results from subsequent *Genetics* cohorts who studied under the language-centred programme.

28. Teaching Evolution as core in First Year Biology

Karen Burke da Silva

School of Biological Sciences, Flinders University, South Australia, 5001, Australia

Most first year Biology courses provide a general overview of the Molecular Aspects of Life and Biological Diversity. Although these courses may be taught from an evolutionary perspective, the theory of evolution itself tends not to be taught until second or often third year level. At this point, many students have moved into streams/majors which do not specifically require evolution as a subject and so only those students who either have an interest in, or for which it is prescribed, go on to study evolution. A survey of first year biology students at Flinders University found that most students enjoyed the Molecular Aspects component of their course. However, student interest in the Biological Diversity course was quite low, with students typically unable to see its relevance to their degree program. The survey also indicated that students gained little understanding of evolution from this course and maintained common misconceptions and misunderstandings. In an effort to improve students' understanding of evolution and to ensure that a greater number of students were exposed to evolutionary theory, I replaced Biological Diversity with Evolution as a course in the first year biology program. Students gave Evolution a much higher evaluation score than Biological Diversity, and far fewer students criticized the relevance of Evolution to their degree program. Interestingly, students' perceptions and misunderstandings did improve after a semester of Evolution; however it did not necessarily change their overall views.

29. The Hands On Genetics and other genetics and evolution computer teaching programs

John Sved

School of Biological Sciences A12, Sydney University, NSW 2006

Hands On Genetics consists of 12 genetics simulation programs that are available as freeware on the internet (http://www.handsongenetics.com) and run under current Windows OS and Macintosh Classic OS. The first program, SimpleGene covers life cycles. There are three classical genetics programs, including the all-purpose Mendelsim that allows the simulation of one, two or three locus crosses, with a variety of associated exercises. There are three molecular biology programs, and other programs including meiosis, inbreeding and pedigrees, and population genetics. The underlying philosophy of the programs is to require students to carry out as much of the work themselves as possible. Examples from several of the programs will be given as well as examples from the Drosim program and two programs for evolution simulation.

30. Systematics and evolutionary history of the most successful Australian geckos – *Gehyra*

Mark J. Sistrom

Evolutionary Biology Unit, South Australian Museum, Adelaide, 5000 Australia and the Australian Center for Evolutionary Biology and Biodiversity, University of Adelaide, 5000 Australia

The geckos of the genus *Gehyra* are distributed Australia-wide, with 17 species currently recognised. Their relatively conservative external appearance masks considerable chromosomal diversity, suggesting that there may be as many as 40 species. My phylogenetic analysis of mitochondrial *ND2* gene sequence data shows that the present species taxonomy of *Gehyra* is indeed inadequate to represent the diversity in the group. However, progress in developing an array of nuclear markers suitable for testing mtDNA phylogenies of closely related squamate reptiles is lagging well behind other vertebrate groups. The ongoing assembly and annotation of the green anole lizard, (*Anolis carolinensis*) genome currently presents an opportunity to identify useful nuclear sequences for the development of a broad range of markers for phylogenetic analysis. The availability of a collection of gecko cDNAs allows a comparison with the green anole genome for the identification of conserved exons bounding introns and as such presents a new opportunity for the development of a comprehensive multilocus marker set. I will present some preliminary data on my efforts to utilize available cDNA and other genomic resources to identify loci useful for intra- and inter-specific phylogenetics and systematics.

31. Phylogeography of the extinct New Zealand moa *Pachyornis*: ancient DNA versus morphology

Nic Rawlence[1], Trevor Worthy[1], Jeremy Austin[1], Alan Cooper[1]

[1] Australian Centre for Ancient DNA, School of Earth and Environmental Science, University of Adelaide.

Geological factors (e.g. volcanism, river formation) and climate change (e.g. glacials and interglacials,

sea level changes) can have significant effects on the phylogeography of a species. Using a combination of ancient DNA and morphology, this talk will discuss how geological and climate change have affected the phylogeography of the three species within the extinct New Zealand moa *Pachyornis*. The results indicate that there is significant phylogeographic structuring within Mappin's moa (*P. geranoides*) and heavy footed moa (*P. elephantopus*), but not crested moa (*P. australis*). The causes of the phylogeographic structuring are hypothesized to be volcanism for *P. geranoides*, and the formation of the Clutha River for *P. elephantopus*. Previously, dispersals of heavy footed moa during the Otiran glaciation (last glacial maximum) from the eastern South Island into North West Nelson/West Coast were thought to have occurred. However, bones provided as evidence of this dispersal have been referred to *P. australis*. The larger individuals previously referred to as *P. elephantopus* in this region are the females of the species, while the specimens originally referred to as *P. australis* are the smaller males. This indicates that this species is sexually dimorphic, something that has not previously been reported. The divergence dates of these speciation and phylogeographic events will be discussed.

32. Patterns of diversification in northern Australian frogs

Price, Luke[1], Donnellan, S.[2] and Mahony, M.[3]

[1]The University of Adelaide, [2]South Australian Museum, [3]University of Newcastle.

Northern Australia has been subject to extensive recurring movements of habitats due to climate change cycles and sea level fluctuations. The complexity of this system has provided substantial challenges to explaining contemporary patterns of species diversification. We aim to explore the effects of historical climate change on patterns of diversification of an anuran assemblage across Northern Australia by comparative phylogeographic analyses of 12 species of the ecologically diverse Australo-Papuan hylid frogs.

Amphibians are well suited to phylogeographic analysis as they experience unique ecological constraints in contrast to other terrestrial vertebrates, e.g. a free-living larva that requires constant access to moisture. These factors affect the ability of taxa to move in response to geological or climate change, and as a consequence the opportunity for range expansion across waterless or saline barriers is limited.

We sequenced the mitochondrial *ND4* gene from individuals sampled across the northern Australian ranges of two species of *Cyclorana* and ten species of *Litoria*. Preliminary analyses indicate congruence of phylogeographic patterns in several species between well-known and newly discovered zoogeographic regions. We discuss whether the observed congruent phylogeographic patterns indicate temporal and spatial positioning of historical biogeographical processes.

33. Water 'Colour' and Fish evolution: A Comparative Phylogeographic Study of Amazonian Fishes across an Ecological Gradient

Georgina M. Cooke[1], Ning L. Chao[2], Luciano B. Beheregaray[1]

[1]Molecular Ecology Lab, Department of Biological Science, Macquarie University, NSW, 2109, gcooke@bio.mq.edu.au and Luciano.Beheregaray@bio.mq.edu.au. [2]Department of Fisheries, Universidade do Amazonas, Brazil, piabas@gmail.com

Amazonia is the largest and the most biodiverse river basin on Earth. It is a complex hydrological landscape that harbors large river systems with dramatic differences in water chemistry. Based largely on optical and sedimentary characteristics, three water types have been distinguished: white, black and clear water. We hypothesize that the diversity of freshwater fish in Amazonia, which is the highest on Earth, has been shaped by a combination of vicariant biogeographic history and an ecological gradient generated by water chemistry. To test this hypothesis, 56 populations of seven co-distributed freshwater fish were sampled in 2005 and 2008 from a vast area encompassing five rivers and the hydrochemical settings of Amazonia. Using a comparative phylogeographic approach, this project will examine consistencies between the evolutionary and distributional histories of populations of each species. Concordant phylogeographic patterns between species will be examined within the context of the geographic histories and hydrological characteristics of the region. Here, using sequence data from two regions of the mtDNA genome, ATP-6 and ATP-8 we present the results for *Steatogeneys elegans* (an electric fish) and *Colomesus asellus* (a puffer fish). *S. elegans* is a "true" freshwater fish, since electric fishes have diversified in freshwater, whereas *C. asellus* is one of the marine-derived fishes thought to have invaded Amazonia during the marine incursions of the Miocene. As such,

phylogeographic comparisons between these taxa should provide interesting temporal perspectives about the mechanisms acting in the diversification of Amazonia fish fauna.

34. Identifying distinct lineages among sympatric Australian alpine *Poa* species to predict evolutionary responses to climate change.

Philippa Griffin

CESAR, Genetics Department, University of Melbourne, 3010 Victoria

Victoria's alpine ecosystems are dominated by the tussock grasses *Poa hiemata* and *P. hothamensis*, with at least six other *Poa* species present. Current identification relies upon highly variable morphological characters, none of which are clearly definitive of species (Vickery 1970). The genus *Poa* is known to be especially prone to polyploidy, apomixis and interspecific hybridisation (Grun 1954). Thus, genetic approaches will help explain and resolve the current taxonomic difficulties. Nuclear microsatellite markers and cpDNA non-coding sequence data were used to investigate genetic variation in herbarium and field specimens of eight Australian alpine *Poa* species. Results provide insights into the reproductive strategies and evolutionary relationships of the different species. This molecular work, combined with drought tolerance experiments, will enable predictions of *Poa* evolution under the warming, drying climate of the Victorian Alps.

Grun P (1954) Cytogenetic studies of *Poa*. I. Chromosome numbers and morphology of interspecific hybrids. *American Journal of Botany* 41, 671-678.

Vickery J (1970) A taxonomic study of the genus *Poa* L. in Australia. *Contributions from the N.S.W. National Herbarium* 4, 145-243.

35. Population genetics analysis reveals substructuring within a population of an Australian reptile tick *Bothriocroton hydrosauri*

Jaro Guzinski

School of Biological Sciences, Flinders University, GPO Box 2100, Adelaide, SA 5001, Australia. Evolutionary Biology Unit, South Australian Museum, Adelaide, SA 5000, Australia

I tested the genetic predictions of a model of population structure that explains a parapatric boundary between two species of ticks that share a large lizard as host in semi-arid mallee woodlands and saltbush shrublands in South Australia. The model is based on alternating areas of variable quality habitat, i.e. 'ridges and troughs', with high and low occupancy rates by ticks respectively. The model predicts relatively low dispersal rates across troughs, which act as 'sinks' and thus the possibility of some genetic differentiation between ridges. Allele frequencies at nine microsatellite loci were sampled from 244 adult *Bothriocroton hydrosauri* ticks (222 males/22 females) collected from 83 lizard hosts found along a transect perpendicular to the parapatric boundary in mid-north South Australia. A plot of tick abundance along the transect showed the ridge and trough pattern predicted by the model. However, the ticks were found to belong to four disparate though syntopic genetic clusters, a structure inconsistent with the predictions of the population model. The clusters exhibited very low levels of gene flow and moderate to high levels of divergence. The barrier to gene flow has not as yet been identified but potentially could involve assortative mating due differences in olfactory cues. I discuss the implications of my findings for speciation in ticks.

36. Conservation genetics of the Bush Stone-curlew (*Burhinus grallarius*)

Robert A. B. Mason[1], Karen-Ann Gray[1], Rebecca N. Johnson[1], Catherine Price[2], Walter E. Boles[1].

[1]Australian Museum, 6 College Street, Sydney, NSW 2010 Australia.[2]National Parks and Wildlife Service, NSW; and School of Biological, Earth and Environmental Sciences, University of New South Wales, Sydney, NSW 2052 Australia.

The Bush Stone-curlew (*Burhinus grallarius*), a ground-dwelling woodland bird, is endemic to Australia and has a wide distribution throughout eastern Australia, where it is speculated to exist as a number of sub-species. Curlew numbers have dramatically declined in abundance in South-eastern Australia due to fox predation and habitat clearance. It is now listed as Endangered in New South Wales and Victoria, and Vulnerable in South Australia. To avoid extinctions of the southern populations (due to small population size, inbreeding and genetic diversity erosion) certain management activities could be necessary. Southern populations could be supplemented either by translocating birds from northern Australia, where the species is still relatively abundant, or through captive breeding. Concern over maintaining the genetic provenance of sub-species, should they exist, has led to this investigation of genetic boundaries. This has implications if decisions are

made to translocate individuals from northern to southern populations or to select candidates for captive breeding programs. To this end, microsatellite loci have been developed and genotyped in individuals from across the geographic range of the Bush-stone curlew. Results, including population genetics and the sub-species question, will be discussed.

37. Where and When Does A Ring Start and End? Testing the Ring Species Hypothesis in a Species Complex of Australian Parrots

Leo Joseph[1,2], Gaynor Dolman[2], Stephen Donnellan[3], Kathleen Saint[3], Mathew Berg[4,5] and Andrew Bennett[4,5]

[1]Department of Ornithology, Academy of Natural Sciences, 1900 Benjamin Franklin Parkway, Philadelphia PA, 19103-1195, USA, [2]Australian National Wildlife Collection, CSIRO Sustainable Ecosystems, GPO Box 284, Canberra, Australia 2601. [3]South Australian Museum, North Terrace, Adelaide, Australia 5000, [4]University of Bristol, School of Biological Sciences, Woodland Road, BS8 1UG, Bristol, UK, [5]School of Life and Environmental Sciences, Deakin University, Geelong, Australia 3217

Speciation despite ongoing gene flow can be studied directly in ring species, which comprise two supposedly reproductively isolated populations connected by a chain of intergrading populations. We applied three tiers of spatio-temporal analysis (phylogeny/biogeography, phylogeography, landscape genetics) to mitochondrial and nuclear data of Australian parrots of the Crimson Rosella *Platycercus elegans* complex to understand the history and present genetic structure of the ring they have long been considered to form. A ring species hypothesis is not a fully adequate explanation of our findings (e.g., discordance in genotypic and phenotypic assignments where terminal differentiates meet). We discuss alternative models involving historical allopatry of populations that were ancestral to the present-day terminal differentiates of the ring. In these models, population expansion shown by population genetics parameters in one of these isolates was accompanied by geographical range expansion, secondary contact and hybridization on eastern and western sides of the ring. Pleistocene landscape and habitat changes then established current distributions and range disjunctions. Populations now show idiosyncratic patterns of selection and drift. We suggest that the history of the species can be thought of as a ring closed on two sides but that selection and drift idiosyncratically drive evolution in different populations within it.

38. Genomic analysis of the multicopied rDNA ITS2 in the mosquito *Anopheles longirostris* Burg (Diptera: Culicidae) and parallel assessment of the mtDNA cytochrome oxidase 1 reveals eight cryptic species in Papua New Guinea

David Alquezar[1], Robert D. Cooper[2], and Nigel W. Beebe[3,4]

[1] Institute for the Biotechnology of Infectious Disease, University of Technology, Sydney. [2]Australian Army Malaria Institute. Brisbane, Australia. [3]School of Integrative Biology, University of Queensland, St Lucia, Queensland, 4072, Australia. [4]CSIRO, Entomology, Indooroopilly, Queensland, 4068, Australia.

In Papua New Guinea (PNG) the mosquito *Anopheles longirostris* has been implicated as a vector of human malaria and appears to exist in a variety of ecological environments, suggesting it may form a complex of genetically distinct but morphologically indistinguishable cryptic species. *Anopheles longirostris* was collected from over 70 sites in PNG, and the multicopy nuclear rDNA ITS2 and mitochondrial DNA cytochrome oxidase 1 (CO1) were assessed for evidence of cryptic taxa. The ITS2 was assessed at three levels –restriction fragment length polymorphism analysis (RFLP: to assess the crude sequence variation), heteroduplex analysis (HDA: to assess the copy variant organisation) and DNA sequencing. Genetic evaluation of over 300 specimens revealed that *An. longirostris* is composed of a minimum of seven ITS2 PCR-RFLP genotypes and eight different HDA genotypes that appear to be evolving independently and show overlapping distributions. The mtDNA CO1 (500bp) was assessed as an independent marker in a Bayesian phylogenetic analysis of 64 individuals representing each of the ITS2 genotypes and revealed eight resolved clades. Phylogenetic comparison of both the nuclear rDNA ITS2 and mitochondrial COI demonstrate areas of strong concordance suggesting the presence of independently evolving cryptic species within *An. longirostris*.

39. Artifactual causes of substitution rate discrepancies

Allen Rodrigo and Peter Tsai

Allan Wilson Centre for Molecular Ecology and Evolution, University of Auckland, Private Bag 92019, Auckland, New Zealand

The phenomenon that has puzzled molecular evolutionary biologists working with ancient DNA is the apparent difference in substitution rates obtained when pairs of sequences diverge at different times. In fact, there is a concave and curvilinear relationship between estimated rates and the times to the most recent common ancestor (MRCA). Authors have suggested biological processes that may account for this curvilinearity, but here we examine whether artifacts of phylogenetic and population genetic inference can account for the observed pattern. In particular, we look at three potential artifacts: (1) a delayed coalescent of lineages in the ancestral population, (2) substitution model misspecification, and/or (3) the censoring of sequence data. We conclude that at least one of these goes some way towards explaining the "L-shaped rates curve".

40. Branch-length estimation bias misleads molecular dating

Matthew J. Phillips

Centre for Macroevolution and Macroecology, School of Botany and Zoology Australian National University, Canberra, ACT 0200, Australia.

Despite recent advances in inferring the time-scale of biological evolution from molecular data, the fundamental question of whether our substitution models are sufficiently well specified to accurately estimate branch-lengths has received little attention. I examine this implicit assumption of all molecular dating methods, on a vertebrate mitochondrial protein-coding dataset. Comparison with analyses in which the data are RY-coded (AG→R; CT→Y) suggests that even rates-across-sites maximum-likelihood greatly under-compensates for multiple substitutions among the standard (ACGT) NT-coded data, which has been subject to greater phylogenetic signal erosion. Accordingly, the fossil record indicates that branch-lengths inferred from the NT-coded data translate into divergence time overestimates when calibrated from deeper in the tree. Intriguingly, RY-coding led to the opposite result. The underlying NT and RY substitution model misspecifications likely relate respectively to "hidden" rate heterogeneity and changes in substitution processes across the tree, for which I provide simulated examples. Given the magnitude of the inferred molecular dating errors, branch-length estimation biases may partly explain current conflicts with some palaeontological dating estimates.

41. Recalibrating the time-scale of human evolution using mitochondrial DNA

Simon Y. W. Ho[1] and Phillip Endicott[2]

[1]School of Botany and Zoology, Australian National University, Canberra ACT 0200, Australia. [2]Magdalen College, University of Oxford, Oxford OX1 4AU, United Kingdom

The time-scale of human evolution, prehistoric migrations, and population changes can be inferred from DNA sequences. The most important aspect of the methodology used to estimate molecular time-frames is the calibration point, the role of which is to convert measures of genetic distances into units of time. Almost invariably, analyses of human evolution have been calibrated with reference to the human-chimpanzee divergence, which is frequently condensed to a point estimate of around 5 Myr despite the existence of considerable uncertainty over this date. Evidence of saturation and selection, however, clearly indicate that this calibration method is inappropriate, and that it is more suitable to use biogeography-based calibrations within the human tree. The impact of calibration choice can clearly be seen when comparing hypotheses of the timing of migration events, such as the colonization of the Americas.

Using a number of candidate calibrations within the human tree, we present a revised time-scale for human evolution based on mitochondrial DNA. We also present estimates of the substitution rate for different portions of the mitochondrial genome, and illustrate some of the complexities of human mitochondrial evolution that are not adequately addressed when using simple models of analysis. Overall, our results call for the use of more sophisticated models of molecular evolution, along with judicious selection of calibration points.

42. The evolutionary rates curve: How accurate are molecular date estimates?

Alan Cooper[1], Jack Da Silva[2]

[1]Australian Centre for Ancient DNA, Earth and Environmental Sciences, University of Adelaide, Adelaide, SA 5005, Australia. Alan.cooper@adelaide.edu.au. [2]Molecular Biomedical Science, University of Adelaide, Adelaide, SA 5005, Australia. Jack.dasilva@adelaide.edu.au

Recent work has indicated that molecular evolutionary rates appear to exhibit time dependency, in that rate estimates vary according to the time period over which they are measured. This is most noticeable in apparently accelerated short-term rate measurements, and can lead to a considerable over-estimate of divergence times when a molecular clock (relaxed or strict) is used to estimate the timing of recent evolutionary events (eg during the past 1-2 Ma). The estimated rates appear to follow a negative exponential curve, with the most rapid estimates resulting from short-term measurements within families or populations, and the slowest from fossil calibrations.

The issue of rate curves falls at the interface of micro- and macro-evolutionary theory, and is partly explained by changes in terminology and methodological assumptions governing research across this boundary. A curve in rates will always be produced if $d > 0$ at $t = 0$, and possible reasons for this phenomena are explored. One key issue appears to be the essential difference between rate estimates below the species level (which are primarily measuring polymorphism within populations) and the much slower rate recorded above the species level (generally calculated with a fossil date) which measure the substitution rate, or mutation rate for neutral loci. The latter is the small proportion of polymorphisms that get actually fixed in a species lineage (*e.g.* by drift, or selective processes) over time. In addition to the effect of polymorphisms, the proportion of saturated sites also appears to play a role in the generation of rate curves.

Several temporally sampled datasets are examined for evidence of rate curves, and different approaches used in an attempt to negate their effect. The results indicate that the problem is widespread, both above and below the species level and that corrections for this behaviour have major impacts on molecular date estimates in the recent past.

43. The head louse, *Pediculus capitis*, has an extraordinary mitochondrial genome

Renfu Shao and Stephen C. Barker.

Parasitology Section, School of Molecular and Microbial Sciences, The University of Queensland, St Lucia, Brisbane, Queensland 4072, Australia.

The mitochondrial (mt) genomes of >1,000 species of bilaterian animals have been sequenced and deposited in Genbank. Without exception, the mitochondrial genomes of these >1,000 species have their 36 or 37 mt genes on a single circular chromosome that is ~16000 bp long. We reported at the 2007 GSA meeting that the body louse, *Pediculus humanus*, has an extraordinarily different mitochondrial genome compared to those of the >1,000 species studied so far. Instead of having all of its mitochondrial genes on a single ~16,000-bp chromosome, the 37 mt genes of *Pediculus humanus* are on 18 mini-circular chromosomes. These mini-circular chromosomes are ~3 kb and have only 1 to 3 genes each. This year we report on the extraordinary mitochondrial genome of the human head louse *Pediculus capitis*.

44. Evolution of expression and function of the prion-protein family genes, *PRNP* and *PRND*

Tatiana Vassilieva, Nagesh Chakka, Jill E. Gready

John Curtin School of Medical Research, Australian National University, Canberra, ACT 0200, Australia

Gene duplication is a major mechanism for evolution of new proteins and functions. Survival of a newly duplicated gene requires acquisition of both a new function and regulatory elements for its expression. *PRNP* and its downstream duplicate, *PRND*, provide a striking example of such an evolutionary process. Both genes are well known in Eutherian mammals, *PRNP*, notoriously for its involvement in neurodegenerative disorders but also for its normal neuroprotective function, and *PRND* as a testis-specific gene whose loss in knockout mice leads to male sterility. Here we report for the first time *PRND* expression in two earlier vertebrate lineages, frog (*Xenopus laevis*) and marsupial (*Monodelphis domestica*). We follow the development of expression – and transcripts – of the new gene using three models: frog, marsupial opossum and mouse. In frog, soon after the duplication, *PRND* relies primarily on regulatory elements of the *PRNP* gene and is expressed in brain as a chimeric transcript, sharing the first two 5' UTR exons of the *PRNP* gene. Development and expression from its own promoter starts initially in

testis and later evolves a broader expression pattern by acquiring new regulatory elements. We see in the three models that during evolution the importance of this more specialized transcript increases and in Eutherian mammals it becomes almost the sole one. Our results support the hypotheses that function evolved, as expected for duplicates, by both sequence and expression divergence.

45. A new model for the evolution of the globin gene clusters in amniotes

Vidushi S. Patel*[,1], Steven J. B. Cooper[2,3], Janine E. Deakin[1], Bob Fulton[4], Tina Graves[4], Wesley C. Warren[4], Richard K. Wilson[4], Jennifer A. M. Graves[1]

*[1]The ARC Centre for Kangaroo Genomics, Research School of Biological Sciences, The Australian National University, Canberra ACT 2601, Australia. [2]Australian Centre for Evolutionary Biology and Biodiversity, The University of Adelaide, South Australia 5005. [3]Evolutionary Biology Unit, South Australian Museum, South Australia 5000. [4]Genome Sequencing Center, Washington University School of Medicine, St. Louis, Missouri 63108, USA.*Corresponding Author:*

vidushi.patel@anu.edu.au

The alpha (α) and beta (β) globin clusters are located together in amphibians and fish, but are on different chromosomes in amniotes (birds and mammals). The discovery of a fossil β-like globin gene (ω) beside the marsupial α-globin cluster suggested that the ancestral α-β cluster duplicated onto two chromosomes, followed by lineage-specific gene loss and duplication, resulting in paralogous α and β clusters in amniotes (Wheeler *et al.,* 2001; 2004). To test this hypothesis, we analysed globin genes and flanking markers from a monotreme mammal, the platypus (*Ornithorhynchus anatinus*). The platypus α-globin cluster (5'-ζ-ζ'-α^D-α^3-α^2-α^1-ω-*GBY*-3') is located on chromosome 21 and the β-globin cluster (5'-ε-β-3') on chromosome 2. Comparison between species revealed that all amniote α clusters are flanked by *MPG*, *C16orf35* and *LUC7L*, as are the α-β clusters of amphibians and fish. However, all amniote β clusters are embedded in an olfactory receptor region. Thus the bird α- and β- clusters are orthologous to the mammalian α- and β- clusters, contradicting previous hypothesis (Wheeler *et al.,* 2001; 2004). We propose a new model where the α- and β- globin clusters evolved from an ancient *MPG-C16orf35*-α-β-*GBY-LUC7L* arrangement. A copy of the original β (represented by ω in marsupials and monotremes) was inserted into an olfactory gene cluster before the amniote radiation (>315 MYA), then duplicated and diverged to form orthologous clusters of β-globin genes in different lineages.

Wheeler, D., R. Hope, S.B. Cooper, G. Dolman, G.C. Webb, C.D. Bottema, A.A. Gooley, M. Goodman, and R.A. Holland. 2001. An orphaned mammalian beta-globin gene of ancient evolutionary origin. *Proc Natl Acad Sci U S A* **98:** 1101-1106.

Wheeler, D., R.M. Hope, S.J. Cooper, A.A. Gooley, and R.A. Holland. 2004. Linkage of the beta-like omega-globin gene to alpha-like globin genes in an Australian marsupial supports the chromosome duplication model for separation of globin gene clusters. *J Mol Evol* **58:** 642-652.

46. Comparative analysis of protein coding genes: Rearrangements and gene gain or loss

Patel, H., Frickey, T., Waters, P., Delbridge, M., and Graves, J.A.M.

Comparative Genomics Group, Research School of Biological Sciences, The Australian National University, Acton, ACT, Australia-2600

Comparative analysis of genomes and proteomes has helped understand fine scale evolutionary mechanisms that elucidate functions and phenotype of organisms. GC rich regions of the genome are also gene dense regions. These regions are proposed to have a high susceptibility to genomic rearrangements. We have analysed protein sequences from 13 vertebrate species using the PhyloGenie suite of tools [1]. Using PhyloGenie, we have been able to define orthologous GC rich regions in 13 vertebrate species. Our preliminary results show that gene-dense regions are highly susceptible to lineage specific deletion and rearrangements. We are examining genomic contents correlated with these deletions, looking particularly into whether break points are frequently reused and the presence of DNA elements that may be facilitating these genomic breaks. This will also aid in understanding the overall DNA conservation of the GC rich regions between these 13 species. This study will add to the delineation of syntenic blocks between these species and provide information about the lineage specific gain and loss of protein coding genes. This will enable us to determine whether these GC rich regions have a high susceptibility to genomic rearrangements.

1. Frickey, T. and Lupas, A. N. - PhyloGenie: automated phylome generation and analysis, Nucl.

Acids Res., 32, pp. 5231-5238 (2004).

47. Telomere length polymorphism in wild Tasmanian devils

Hannah S. Bender[1,2], Elizabeth P. Murchison[2], Margaret A. Strong[3], Daniel A. McMillan[1], Tariq Ezaz[1], Carol W. Greider[3], Anne-Maree Pearse[4], Gregory J. Hannon[2] and Jennifer A. Marshall Graves[1]

[1]Research School of Biological Sciences, The Australian National University Canberra, ACT 2601, Australia. [2]Watson School of Biological Sciences, Cold Spring Harbor Laboratory, Cold Spring Harbor, NY 11725, USA. [3]Department 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

Devil Facial Tumour Disease (DFTD), an aggressive transmissible tumour is rapidly spreading through the wild Tasmanian devil population, threatening east coast populations with extinction and encroaching on the, until now, healthy western Tasmanian devils. Cytogenetic research suggests that DFTD is caused by a transmissible cell line. The tumour karyotype is highly rearranged with several chromosomes, including the sex chromosomes, apparently missing and with the addition of four unidentified marker chromosomes. This karyotype is found in all tumours, regardless of stage of tumour progression or the sex of the animal. In order to investigate the telomere length in this remarkably stable tumour, we characterized chromosomes of 2 male and 3 females from different locations in Tasmania, using Fluorescent in situ hybridisation and telomere restriction fragment analysis. Surprisingly we found that all devils possess two very different telomere lengths (50-70kb and approximately 200kb), and were evidently heterozygous for long and short telomeres. This unusual telomere distribution could be the result either of recent hybridization of two devil populations, or of a parent-specific telomere modification.

48. Genetics and ecogeographical studies in subclover (Trifolium subterraneum)

Kioumars Ghamkhar[1], Megan Ryan[2], Phil Nichols[3], Richard Snowball[3], Rudi Appels[4]

[1]Centre for Legumes in Mediterranean Agriculture, Faculty of Natural and Agricultural Sciences, The University of Western Australia, Western Australia, 6009 Australia. School of Plant Biology, Faculty of Natural and Agricultural Sciences, The University of Western Australia, Western Australia, 6009 Australia. [3]Department of Agriculture and Food Western Australia, 3 Baron-Hay Court, South Perth, Western Australia, 6151, Australia. [4]Centre for Comparative Genomics, Murdoch University, Western Australia, Australia

Flowering time, hardseededness, leaf marks, and several other agro-morphological traits of *Trifolum subterraneum* are being studied for any linkage with microsatellite markers using QTL mapping. On another front, overall genetic diversity is being screened using germplasm location, rainfall, temperature, agronomic, morphological, and molecular (fAFLP) data. As a result of using core collection development techniques, the size of a germplasm collection of 10,000 accessions and genotypes will be reduced to about 300 for future selection and plant breeding prgrams. The preliminary linkage studies suggest that hardseededness (a very significant character in *T. subterraneum*) is controlled by multiple genes. Some significant relationships have been also found among several agromorphological data.

49. The role of the Major Histocompatibility Complex in Devil Facial Tumour Disease

Katherine Belov, Hannah Siddle, Jolanta Marzec, Claire Sanderson.

Faculty of Veterinary Science, University of Sydney, NSW 2006, Australia

Tasmanian devil facial tumour disease is a transmissible cancer. The disease is decimating devil numbers and may lead to extinction of the species in the wild. The Major Histocompatibility Complex (MHC) is the most important region of the genome in terms of disease resistance and graft rejection. Genes within this region are considered the most polymorphic in vertebrates. We have found that Tasmanian devils lack diversity at the MHC, and have suggested that this paucity in MHC diversity has enabled the spread of DFTD (Siddle et al. 2007). Recently, we have identified MHC-disparate animals in isolated devil populations. I will present details about the alleles identified in MHC-disparate animals and discuss the importance of these animals for ongoing studies into disease resistance. I will also discuss efforts to incorporate MHC data into the captive breeding program.

Siddle, H. V., Kreiss, A., Eldridge, M. D., Noonan, E., Clarke, C. J., Pyecroft, S., Woods, G. M., and Belov, K.: Transmission of a fatal clonal tumor by biting occurs due to depleted MHC diversity in a

threatened carnivorous marsupial. *Proc Natl Acad Sci U S A*, 2007

50. The expression, evolution and role of ligand-gated chloride channels in insecticide resistance

Emily Remnant, Phil Batterham and Phillip Daborn

Centre for Environmental Stress and Adaptation Research (CESAR), Department of Genetics, Bio21 Molecular Biotechnology Institute, The University of Melbourne

The major inhibitory neural receptors of the insect nervous system are the ligand-gated chloride channels (LGCCs). This gene family contains a formidable set of highly effective neural insecticide targets for a number of chemical classes, including dieldrin, fipronil and avermectin. The genome of *Drosophila melanogaster* contains 12 LGCC subunits. These genes are being studied from three different perspectives; expression, evolution and insecticide resistance.

Three of the subunits are highly diverged and vary in copy number across insect species. RT-PCR and *in situ* hybridisation have revealed unexpected expression of these genes in peripheral tissues such as ovaries and the gut.

Heterozygous null mutants of the subunit gene *Rdl*, which is a known target of phenylpyrazoles, show increased sensitivity to phenylpyrazoles. Based on this information, RNAi of the LGCC genes will be used to investigate the function of these genes in neural and peripheral tissues, and with EMS mutagenesis, examine their potential role as candidate targets for phenylpyrazoles and other insecticides.

The Sir Ronald Fisher Lecture

51. The impact of ancient polyploidisations on eukaryotic genome evolution

Ken Wolfe

Smurfit Institute of Genetics, Trinity College Dublin, Ireland

Polyploidisation is a natural phenomenon whereby the DNA content of an organism becomes doubled. It occurs most frequently by the production of gametes that did not go through meiosis. Many examples of recently-formed polyploid species are known, such as tetraploid salmon or hexaploid bread wheat. Genome sequencing projects have revealed that many older events of polyploidisation (also called whole genome duplication or WGD) occurred during the evolution of eukaryotes. Indeed, rather few sequenced eukaryotes have turned out not to show evidence of WGD.

WGD has many interesting evolutionary consequences, which I will discuss in the context of our studies on the genomes of yeast species. We are fortunate to have the complete genome sequences of several yeast species (including Saccharomyces cerevisiae) that are descended from a common ancestor that underwent WGD, as well as several outgroup species that branched off shortly before this WGD event happened. Comparing the order of genes along their chromosomes, which is surprisingly well conserved, allows us to study the genome-evolutionary events that occurred in the wake of the WGD. We find that many extra copies of genes were unnecessary and were deleted from the genome very rapidly. This process of gene loss is likely to have caused new species to be formed, because genomes that lose different copies of duplicated genes will quickly become reproductively isolated from one another. We find that among the gene pairs that have survived in duplicate after the WGD, the rate of sequence evolution has been both accelerated and asymmetrical. Many of the surviving gene pairs consist of one higher-expressed slower-evolving member, and one lower-expressed faster-evolving and often dispensable member; in some instances the faster-evolving gene has gained a new function. Lastly, by studying the pattern of gene loss after WGD, we have discovered a bizarre process of DNA erosion from the regions flanking the mating-type locus in yeast.

Ken Wolfe is Professor of Genome Evolution at the Smurfit Institute of Genetics, Trinity College Dublin (Ireland). His current research interests are on the evolution of eukaryotic genome organization, particularly relating to how new genes are formed and how the order of genes along chromosomes can evolve. His laboratory formed part of the international consortia that sequenced and analyzed the Saccharomyces cerevisiae genome (1996) and the human genome (2001), and with Denis Shields in 1997 he discovered an ancient whole-genome duplication during the evolutuion of S. cerevisiae. Prof. Wolfe is an Associate Editor of Molecular Biology and Evolution and an Academic Editor of PLOS Biology, and a Fellow of the American Association for the Advancement of Science. The Wolfe laboratory runs the "PubCrawler" literature alerting internet service which has over 30,000 users worldwide. (www.pubcrawler.ie).

Placental mammals 100 - 105 myr

Birds reptiles 310

Marsupials 147

Monotremes 168 Skeleton reptile-like

Tammar wallaby 3.5gb Schmaans 2x WGS seq.

Xist no counterpart in marsupials/frog/chicken

X-inactivation occurs

Platypus 2.6 GB. 6x WGS assembly

52 chr

10 sex chr

milk genes

egg yolk genes

Human 1340 genes on X SRY

45 or Y unique active TDF ⇒ testis

Not ZFY autosomal in marsupial

Marsupials - no homology w X don't pair

Birds ZW ♀ DMRT1 ZZ ♂ 2 DMRT1 z = human 9.

XY Chicks 1, 4

Snake ZZ/ZW ≠ bird Z

Dragons ZZ/ZW ♀ chr + Hot ⇒ ♀

Turtles temp opp dir to crocodiles

└ Z = chr 5 in turtle.

XY - 145 myr - term in marsup.

monotremes 5 Xs & 5 Ys Nature 2004 Frank Grutzner

Ancient X genes here chr 6 platypus.

Platypus - sex chr like chicken Z

Why do so many X-linked syndromes combine mental retardation & gonadal abnormality?

SOX3 - brain dev & testis differentiation?

mole voles no Y, no SRY

52. Weird animal genomes and sex

Jennifer A. Marshall Graves

Research School of Biological Sciences, The Australian National University, Canberra ACT 2601.
Email jenny.graves@anu.edu.au

Our strategy is to compare the chromosomes, genes and DNA between the most distantly related mammals, and even with birds and reptiles. The genomes of Australia's unique kangaroos and platypus, now being completely sequenced, are particularly valuable because these "alternative mammals" are not too close to find informative differences from human, nor too distant to compare DNA sequences.

In humans and other mammals, females have two X chromosomes, and males a single X and a Y. The *SRY* gene on the Y switches on the development of testis, which pumps out male hormones and masculinizes the embryo. The human X is a mid-sized, ordinary sort of chromosome, though it has more than its fair share of genes involved in male sex and reproduction (and maybe sexual behaviour and intelligence), and it is inactivated in females by epigenetic silencing. But the Y is a genetic wasteland – small and repetitive, bearing only 45 genes, most active only in testis. Birds and reptiles have completely different triggers for sex determination; either genes on unrelated sex chromosomes (Z and W), or environmental triggers like temperature. Or both, as we recently discovered in dragon lizards.

How did human sex chromosomes get to be so weird? Kangaroo sex chromosomes give us clues about the original mammal sex chromosomes, and the evolution of X inactivation. The bizarre multiple platypus sex chromosomes turn out to be completely unrelated to the XY of other mammals, instead showing homology to the bird ZW. Instead, the XY of other mammals is homologous to an ordinary autosome (platypus chromosome 6). Human sex chromosomes, then, are relatively young.

Thus comparing X and Y chromosomes between distantly related mammals, as well as their antecedents in birds and reptiles, show that the human X and Y evolved from an ordinary chromosome pair only 166 million years ago. Since then, the Y degraded progressively; at a rate that will see it completely disappear in 5 or 6 million years.

Jennifer Marshall Graves was born and educated in Adelaide. She received a BSc (Hons) and MSc from Adelaide University, then a PhD in Molecular Biology at the University of California at Berkeley in 1971. She lectured at La Trobe University for nearly thirty years then moved to ANU in 2001. Jenny now heads the Comparative Genomics Research group in the Research School of Biological Sciences at ANU, and directs the ARC Centre of Excellence for Kangaroo Genomics. She has produced three books and 350 research articles. She is a Fellow (and Foreign Secretary) of the Australian Academy of Science and 2006 L'Oreal-UNESCO Laureate. She has received a number of honours and awards, including the Macfarlane Burnet medal in 2006.
Jenny works on animal genetics and genomics. Her group uses the distant relationship of Australian mammals and other vertebrates from humans to understand how genes and chromosomes evolved and how they work in all animals including humans. Her laboratory uses this unique perspective to explore the origin, function and (dismal) fate of human sex chromosomes.

53. Lower metazoan genomes-a window on genome evolution

Eldon E. Ball[1], David J. Miller[2]

[1] Centre for Molecular Genetics of Development and Research School of Biological Sciences, ANU, Canberra. [2]ARC Centre of Excellence for Coral Reef Studies and Comparative Genomics Centre, JCU, Townsville

Genome sequences and ESTs from several lower metazoans have recently become available, including those of the sponge, *Amphimedon*, the placozoan, *Trichoplax*, and the cnidarians, *Nematostella* , *Hydra* and *Acropora.* Comparison of these genomes can provide a window into how the genomes of bilaterians evolved and has yielded some generalities as well as some surprises. Among the latter for the Cnidaria are the complexity of their genomes, the presence of apparently ancient 'non-metazoan" genes and the occurrence of all developmental pathways found in higher animals. I will illustrate the expression of representatives of these pathways from our work on axis determining genes in the coral, and discuss changes in the roles of TGFbs and Hox genes during animal evolution. Finally, I will discuss the galaxins, which are expressed in the organic skeleton of *Acropora*, as an example of a thus-far coral specific gene family.

54. An integrated approach to undertaking diverse studies in comparative genomics

Matthew Bellgard

Centre for Comparative Genomics, Murdoch University, Western Australia.

The Centre for Comparative Genomics, a Western Australian Centre of Excellence, is undertaking a number of collaborative research and development projects that generate a range of molecular

Frank Grützner

genome organisation in spp — use large-scale genome tissues

mammals — non-random organisation — platypus?

Chicken — random? — microchromosomes — Z random — many genes

Platypus — many chr some dot-like — IX IX medial rest metacentric in sp.

½ of BACS had non-random org in chicken

microchroms much less compacted in chicken

Janine Deakin

14 BACS — 19 genes expressed in fibroblasts
+ 2 autosomal controls. 9 pseudoautosomal
HPRT G6PD 7/9. 1:1 2? not detected

X sp. high variation but most seem to have some level dosage compensation

Imprinted X-inactivation? 3 genes tested all bi-allelic expression
ie not imprinted

RNA fish for primary transcript for trans silencing
pseudoautosomal genes only. 1 trans signal for some

Sex sp. — partial silencing. But not coordinated?

sequence data. Sequencing projects include: whole genomes of three closely related pathogenic bacterial species; three Megabases of Wheat in a region of agronomic importance to Australia; and small RNA sequencing of the cattle tick at different developmental stages to identify and functionally characterise microRNAs.

The Centre is also involved in a diverse range of other collaborative comparative genomic, functional genomic and proteomic studies. In all of these projects, Bioinformatics, high resolution visualisation, software development statistical analysis, and high performance computing are tools essential for the integration and interpretation of data. In this regard, the Centre is also engaged in tool development.

In this presentation, I will provide highlights of the Centre's comparative genomic activities as well as showcase some of the innovative bioinformatics strategies the Centre is implementing in order to conduct this analysis in a coordinated and efficient manner.

55. Large-insert genomic clones – a new tool to study genome organisation in sperm

Frank Grützner and Enkhjargal Tsend-Ayush

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

A large body of evidence shows that chromosomes are non-randomly organised in mammalian sperm. Whole-chromosome painting probes have been widely used to identify the position of chromosomes in sperm of various species. In addition centromere and telomere repeats revealed new aspects of genome organisation in sperm (e.g. chromocenters and chromosome looping).

Genome projects from a growing number of vertebrate species offer a good opportunity to obtain defined genomic clones from regions of interest. To get a more accurate picture of genome organisation, we mapped chromosome specific BAC clones in chicken and platypus sperm, which are very similar in shape and yet previous data showed very different organisation of chromosomes in the nucleus.

BAC clones from different autosomes and sex chromosome in platypus and chicken revealed non-random organisation in almost half of the chromosomes in chicken and almost all chromosomes in platypus. The use of the BAC clones allowed us to investigate orientation and regional condensation. Here we demonstrated that chromosomes maintain orientation in sperm and show large differences in condensation. Our results suggest that BAC clones are good tools to investigate genome organisation in sperm. In addition our data reveal new aspects of genome organisation and evolution in platypus and chicken sperm.

56. The status of dosage compensation in the multiple X chromosomes of the platypus

Janine E. Deakin, Timothy A. Hore, Edda Koina, Jennifer A. Marshall Graves

Research School of Biological Sciences, The Australian National University, Canberra ACT 0200, Australia

Dosage compensation equalizes the expression of genes found on sex chromosomes so that they are equally expressed in females and males. In eutherian and marsupial mammals, this is accomplished by silencing one X chromosome in females. In birds, dosage compensation is variable and incomplete. Whether dosage compensation exists in the third group of mammals, the egg-laying monotremes, is of considerable interest, particularly since the platypus has a complex sex chromosome system in which five X and five Y chromosomes share considerable homology with the chicken Z. As part of the platypus genome project, genes have now been assigned to four X chromosomes. We have shown by qRT-PCR that there is some evidence for dosage compensation but it is variable between genes. SNP analysis of several X-borne genes showed that both alleles are transcribed in a heterozygous female, so dosage compensation in platypus is not via imprinted X-inactivation as it is in marsupials. However, RNA-FISH shows that there is a difference in the probability of expression for X-specific genes, with about 50% of female cells having two active copies of an X-gene while the remainder have only one. This means that, although the platypus has the variable compensation characteristic of birds, it also has some level of inactivation characteristic of dosage compensation in other mammals.

57. From immunome to venome: Convergent evolution of platypus and reptile venom genes

Camilla M. Whittington[1], Anthony T. Papenfuss[2], Katherine Belov[1]

[1]Faculty of Veterinary Science, University of Sydney, NSW 2006. [2]The Walter and Eliza Hall Institute of Medical Research, Parkville, VIC 3050

The recent publication of the platypus genome

Camilla Whittington.

Defensins. Pores in bact. walls. 3 S-S bonds
7 β-def.
4 α-def.

β-def. homology to genes coding for Platypus venom most abundant amongst 19 peptides
3 adj genes on an X

dupl. → 55 m – echidna Lead venom (has spur remnant)
similar to cootamines & CLP in reptiles
also evolved from defensins.
but from a diff lineage.

vDLP are expressed in other tissues
one are in ♀ (spleen?).
most recent is ~~xenorop.~~ specific.
Are they antimicrobials?
immune function

(Warren *et al.*, in press) offers a unique opportunity for the study of one of the most interesting features of the platypus: its venom. Male platypuses possess spurs that are used as offensive and defensive weapons to deliver a complex mixture of venom peptides into the victim. Our knowledge of platypus venom is incomplete. It is known that the venom contains peptide fractions like defensin-like peptides (OvDLPs), but their function is not known and there are many more unidentified fractions. Considering the range of useful molecules that have been identified in snake venom, it is hoped that research into the pharmacology of platypus venom may yield novel drugs and new targets for painkillers.

Working as part of the platypus genome sequence annotation team, we have characterised some of the venom genes of the platypus. Interestingly, we have found that OvDLPs have evolved through duplication from existing antimicrobial genes, as have some components of snake and lizard venom, in a compelling example of convergent evolution (Whittington *et al.*, 2008). We have shown that the OvDLPs arose prior to the divergence of platypus and echidnas, suggesting that the ancestral monotreme was venomous. In an attempt to determine the function of the OvDLPs, we have carried out expression assays, and have found that they are distributed in a wide range of tissues, suggesting a broader role than previously suspected.

Warren, W., Hillier, L., Marshall Graves, J., Birney, E., Ponting, C., Grützner, F., Belov, K., Miller, W., Clarke, L., Chinwalla, A., Yang, S., Heger, A., Locke, D., Miethke, P., Waters, P., Veyrunes, F., Fulton, L., Graves, T., Puente, X., López-Otín, C., Ordóñez, G., Eichler, E., Deakin, J., Thompson, K., Kirby, P., Papenfuss, A., Wakefield, M., Olender, T., Lancet, D., Huttley, G., Smit, A., Renfree, M., Pask, A., Temple-Smith, P., Batzer, M., Walker, J., Konkel, M., Harris, R., Taylor, J., Whittington, C.M., Gemmell, N., Buschiazzo, E., Jentzsch, I., Merkel, A., Schmitz, J., Zemann, A., Churakov, G., Kriegs, J., Brosius, J., Murchison, E., Sachidanandam, R., Smith, C., Stark, A., Hannon, G., Rens, W., Ferguson-Smith, M., Lefèvre, C., Sharp, J., Nicholas, K., Ray, D., Kube, M., Reinhard, R., Pringle, T., Flicek, P., Hardison, R., Center, W.U.G.S., Mardis, E.Wilson, R., In press. Genome analysis of the platypus reveals unique signatures of evolution. Nature

Whittington, C.M., Papenfuss, A.T., Bansal, P., Torres, A.M., Wong, E.S.W., Deakin, J.E., Graves, T., Alsop, A., Schatzkamer, K., Kremitzki, C., Ponting, C.P., Temple-Smith, P., Warren, W.C., Kuchel, P.W.Belov, K., 2008. Defensins and the convergent evolution of platypus and reptile venom genes. Genome Res. 18

58. The evolution of resistance to bacterial infection in *Drosophila melanogaster*

Yixin Ye and Elizabeth A. McGraw

School of Integrative Biology, The University of Queensland, Queensland, 4072, Australia

Drosophila harbour substantial amounts of genetic variation for antibacterial immunocompetence. Investment in immunity is believed to be traded off against costly fitness-related traits including development, growth, maintenance and reproduction. To understand the way in which insects invest in fighting bacterial infection, we allowed wild-caught *Drosophila melanogaster* to evolve resistance in response to systemic infection with *Pseudomonas aeruginosa* over 10 generations. *P. aeruginosa* is a Gram-negative bacterium virulent not only to plants and insects, but is also one of the top three causes of opportunistic infection to humans. In response to selection, resistance to infection increased from 15% to 70%. The increase in resistance was costly for flies as evidenced by reduced larval viability, longevity, and female productivity and a rapid loss of resistance once selection was removed. Counter to expectation we also saw increased developmental rates in the resistant flies. Lastly, we performed genome wide transcriptional profiling to compare lines selected for resistance to controls to identify candidate genes in the host that have implications in antimicrobial defense and the evolution of resistance.

59. Truncated Presenilin peptides with potent dominant negative activity

Michael Lardelli

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

The presenilin genes, *PSEN1* and *PSEN2*, are loci for the majority of characterized mutations causing Familial Alzheimer's Disease (FAD). Over 160 dominant, missense mutations causing FAD are known in *PSEN1* but almost no mutations causing protein truncation. This led to the assumption that the latter are null. By injecting morpholino antisense oligonucleotides into zebrafish embryos we disrupted splicing of transcripts from the zebrafish *psen1* gene. This causes a strong dominant negative phenotype when disruption occurs in the region of exon 7. We confirmed that this was due to production of truncated peptides by injection of engineered mRNAs. Using our unique bioassay for *psen2* activity we found that truncated Psen1

Tom Harrop.

Gal4 line. UAS line

P450 CPR Golgi. } here are other redox ptnes.
Dare mito chondrion

Drivers - use for RNAi
w tubulin → lethality

5' HR driver fat body + midgut expressed
non - lethality

Dare - P4 mit. P450s

Jeremy Shearer
boxes 70% homozygous

Nimble Gen sequence capture array

peptides also interfere with *psen2* activity. We present a model for dominant negative action of truncated Psen1 peptides and discuss possible implications for the etiology of sporadic Alzheimer's disease.

Nornes, S, Newman, M, Verdile, G, Wells, S, Stoick-Cooper, C, Tucker, B,

Frederich-Sleptsova, I, Martins, R, Lardelli, M (2008) Interference with splicing of Presenilin transcripts has potent dominant negative effects on Presenilin activity. *Human Molecular Genetics*, 17: 402-412

60. Targets of the CreB regulatory deubiquitinating enzyme in *A. nidulans*.

Kamlangdee, N, Lockington, RA, and Kelly, JM

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

The creB gene of A. nidulans encodes a regulatory deubiquitinating enzyme, and mutations in creB cause pleiotropic effects on carbon and nitrogen metabolism. Strains lacking CreB grow poorly on some media, such as medium containing quinic acid as a carbon source, and cause deregulation of some genes usually regulated by carbon catabolite repression. Based on the phenotype of creB mutations, we have taken a candidate protein approach to investigate whether the quinate ion transporter, QutD, or the carbon catabolite repressor, CreA, are targets of the CreB deubiquitinating enzyme. Results will be presented from experiments in which strains containing epitope-tagged versions of the CreB, QutD and CreA proteins have been used to determine the ubiquitination status of QutD and CreA, whether they co-immunoprecipitate with CreB, and whether the amount of each protein is dependent on the creB status of the cell. These experiments indicate that QutD, and a fraction of CreA, are ubiquitinated proteins, and targets of the CreB deubiquitinating enzyme.

61. Assessment of Cytochrome P450 function in *Drosophila melanogaster* by RNA interference of the P450 redox partners CPR and Dare.

Tom Harrop, Philip Batterham and Phillip J. Daborn

Centre for Environmental Stress and Adaptation Research (CESAR), Department of Genetics, Bio21 Molecular and Biotechnology Institute, The University of Melbourne, Victoria, 3010, Australia

Cytochrome P450s are a large superfamily of genes that are found in all eukaryotic organisms. In insects, the enzymes encoded by P450 genes are involved in many different functions but despite their importance in a range of biological processes, the role of most of the 85 P450s in *Drosophila melanogaster* is not known. A new system for investigating P450 functions *in vivo* has been developed. Two of the redox partners of P450 proteins, CPR and Dare, supply electrons to the P450 during the reaction cycle and are essential for P450 function. RNAi of *Cpr* and *Dare* is being used with the *GAL4/UAS* system to reduce P450 activity in specific tissues and life stages. For example, reduction of P450 activity in the detoxification tissues can be used to assess the potential for P450-mediated insecticide resistance. This system will also be used to discover novel P450 functions.

62. Using the canine SNP array to identify disease genes

Jeremy R. Shearman and 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 show winning males siring a large proportion of the next generation. This can result in recessive mutations, which are identical-by-descent, becoming common in a breed. Affected dogs would thus be homozygous for a large region surrounding the disease gene due to the extensive linkage disequilibrium in domestic dog breeds. This makes dogs a good model for identifying the genetic causes of disease. We have successfully used the candidate gene approach and microsatellites to identify mutations responsible for 2 recessive diseases. The recent development of the Affymetrix canine SNP array allows whole genome association studies to identify the disease region using a small number of unrelated affecteds and controls. This will simplify the process of identifying disease genes. We are using the SNP arrays to search for the gene that causes an ataxia in the Australian Kelpie by identifying a large region of shared homozygosity in affecteds. This will allow a test for carriers to be developed and provide a model for human ataxia.

63. Molecular genetics of beef fat colour

Rugang Tian[1], Wayne Pitchford[1], Cynthia D.K.Bottema[1].

[1]Discipline of Agricultural and Animal Science, The University of Adelaide, Adelaide, South Australia, 5371.

Fat colour is one of the most important beef quality criteria. Beef with yellow fat cost Australian cattle producers approximately $18 million annually as very yellow carcasses are rejected from export markets. Yellow fat is caused by the accumulation of ß-carotene in adipose tissue, and this deposition appears to be under genetic control. The objectives of this project are to identify genes controlling beef fat colour and determine the mode of inheritance. Candidate genes were selected based on QTL mapping data and their roles in ß-carotene metabolism. These candidate genes were sequenced to find single nucleotide polymorphisms (SNPs). The SNPs were then genotyped to verify that these candidate genes affect fat colour. A total of 12 SNPs for the BCDO2 gene were identified including one functional SNP, which creates a stop codon. A total of 10 SNPs were found in the RDHE2 gene with 3 SNPs resulting in amino acid substitutions. Association studies indicate that both BCDO2 and RDHE2 affect fat colour and have epistatic effects.

64. The *Drosophila* Protein Tyrosine Phosphatase dPTP61F regulates insulin signalling and growth

Bree Buszard[1], Tony Tiganis[2], and Coral Warr[1]

1. School of Biological Sciences, Monash University 2.Department of Biochemistry and Molecular Biology, Monash University.

Protein tyrosine phosphatases are key regulatory enzymes of tyrosine phosphorylation-dependent signalling, involved in virtually all aspects of growth and development. dPTP61F is a *Drosophila* protein tyrosine phosphatase that has been shown to negatively regulate JAK/STAT signalling and thus may be involved in processes such as inflammation, tumorigenesis and development. The mammalian homologs of dPTP61F also negatively regulate insulin signalling, however dPTP61F had not previously been shown to have this function. dPTP61F has 4 isoforms that differ in their subcellular localisations and N and C terminal domains, and we are interested in how these differences may lead to differential regulation of signalling pathways.

To further characterise dPTP61F we have used imprecise excision of a P-element to create a deletion mutant that removes 2 of the isoforms. The mutant is homozygous viable but has fertility defects, and we are investigating its effect on several signalling pathways. We are also specifically investigating the effect of the 4 isoforms on insulin signalling using a combination of tissue-specific misexpression and RNA interference experiments, as well as epistasis experiments with mutations in the insulin signalling pathway. These experiments show that dPTP61F negatively regulates insulin signalling in an isoform specific manner.

64a. Meiotic sex chromosome inactivation in monotremes

Tasman Daish, Frank Grützner and Enkhjargal Tsend-Ayush.

Discipline of Genetics, School of Molecular and Biomedical Science, The University of Adelaide, South Australia.

In mammals, pairing of sex chromosomes at male meiosis is restricted with the unpaired regions being subject to silencing during prophase 1. This silencing is achieved by the accumulation of specific histone variants and histone modifications leading to the formation of the heterochromatic sex body. Monotremes have a unique and complex sex chromosome system involving the formation of a meiotic sex chromosome chain however it remains unknown whether any or all of the sex chromosomes undergo meiotic silencing.

Hallmarks of meiotic silencing include accumulation of the histone variant mH2A and phosphorylation of H2AX. Platypus male meiotic cells immunostain for mH2A adjacent the nucleolus organising region. Using sex chromosome-specific BAC clones for FISH probes, we observed some sex chromosomes in the chain to be excluded from the mH2A enriched regions suggesting differential silencing of sex chromosomes may occur.

Phosphorylation of H2AX (gH2AX) is a rapid and highly conserved response to double strand break (DSB) formation and also accompanies meiotic sex chromosome silencing however we could not detect this modification in platypus testis histone extracts. However, we could induce gH2AX formation by irradiating platypus fibroblasts but this response was significantly reduced and delayed compared to that observed in mouse fibroblasts.

We conclude there to be differential sex chromosome silencing occurring in platypus and that monotremes may not have a conserved DSB

Brian Dalrymple

1.5 - 2 m 18 walks 60K SNP chip
6 animals low quality
60 20X reduced representation (Restr. Enz 1-5% genome)
high quality

SOLEXA Sequence → 30 odd bases
of ends
BACs check arrangement (tail to tail) 33-35
~~Not~~ remove rearranged. Soap.

2.5m 454 contigs ~480bp length
~43% genome Sanger > Solexa > 454
30% unique genome

Dave Adelson

LINES 3-6 kb fully ref. Pol II promoter 40-50% of mammalian genome
SINES Pol III promoter

Multiple alignments TALS need to mask
Cluster then align & PILER repeats
use Muscle

Repeat Scout
Cluster (BLAST CLUST)
End up with repeat library → repeatmasker to mask genome

Most other mammals have same lines, cow has one more of its own BovB/LINE (non-LTR)
↑
opossum has it too
believed to be horizontally transmitted from snakes (squamates)
marsupial monotremes

Gavin Huttley

Don't use PAML.
Markov codon subst model.
MG muse & Gaut 1994
or Goldman & Yang. 1994

response mechanism as part of the meiotic sex chromosome silencing pathway.

65. Building the sheep genome using comparative genomics and new generation sequencing technologies.

Brian. P. Dalrymple on behalf of the International Sheep Genomics Consortium

CSIRO Livestock Industries, Queensland Bioscience Precinct, St Lucia, Queensland 4067, Australia.

Researchers now expect access to the assembled and annotated genome sequence of their favourite organism. However, the short length of the sequence reads and the large amount of data generated by the new faster genome-sequencing technologies creates significant new challenges for the bioinformatics community, especially in putting all of the information together into an accurate assembly of a mammalian genome. Using 454 sequencing, genomic sequence has been obtained from six sheep each of a different breed by Baylor College of Medicine and AgResearch (NZ). Approximately 3-fold coverage of the "sheep" genome was obtained, allowing only very short contigs to be constructed. As the primary focus of the project was to identify SNPs, only a small number of paired-end reads were obtained, thus severely limiting genome assembly based solely on sheep sequence. However, by using the bovine genome assembly and comparative genomics we have reordered the bovine genome sequence into our best estimate of the sheep genome structure (a "virtual sheep genome"). Then, by replacing the bovine sequence with the relevant sheep sequence, we have constructed a true sheep genome assembly. The few available paired-end read sequences were then positioned onto the assembly and showed a high congruency, suggesting that the assembly method was successful.

66. Identification and annotation of repeats in the bovine genome.

David L Adelson[1], Joy M Raison[1] , Robert C Edgar[2]

[1]The University of Adelaide, Adelaide, SA, Australia
[2]Tiburon, CA, U.S.A.

We have created a pipeline to identify and annotate DNA repeats in the bovine genome, using two pre-existing tools PALS/PILER and RepeatScout) which had previously not been used on an entire mammalian genome. The pipeline breaks up the genome into manageable chunks to run PALS in a parallelized fashion on a computer cluster. The chunks are then concatenated at the chromosome level and used as input for PILER, generating clustered, consensus sequences for repeats on each chromosome. RepeatScout was run on individual chromosomes and its output converted to make it compatible with PILER output. Redundancy was minimized by genome wide clustering of the consensus sequences, along with the RepeatScout output, by using BLASTCLUST to generate globally alignable, non-redundant consensus sequences. In this fashion we have identified many previously known repeats and a number of new repeats. Correlation analysis of repeat coordinates across the genome has identified some repeat types as being spatially correlated. These correlations are being explored in order to identify potential molecular mechanisms that could bias or determine repeat insertion sites. Comparative analysis of the bovine repeat landscape with respect to other mammalian genomes reveals both conserved and novel repeat profiles.

67. Context and Codons: standard models are biased by sequence composition

Gavin Huttley[1] and Von Bing Yap[2]

[1]John Curtin School of Medical Research, The Australian National University, Canberra ACT 0200 Australia; [2] Department of Statistics and Applied Probability National University of Singapore

Codon models of sequence evolution are central to comparative approaches to testing the Neutral theory of molecular evolution for protein coding genes. The most popular of these are continuous time Markov process models that derive from the work of Goldman and Yang (1994, hereafter GY) and Muse and Gaut (1994, hereafter MG). Evidence has been presented that estimates of the ratio of nonsynonymous to synonymous substitution are different between the GY and MG models in a manner affected by sequence composition. For related families of context dependent models, we will demonstrate that in fact it is the MG model class that are more robust to the influence of sequence composition. Both families of models are implemented in the development version of the evolutionary modeling toolkit PyCogent (Knight et al, 2007).

N. Goldman, Z. Yang. *Mol. Biol. Evol.*, 11:725–36, 1994.
R. Knight, et al. *Genome Biol*, 8:R171, 2007.
S. Muse, B. Gaut. *Mol. Biol. Evol.*, 11:715–24, 1994.

68. Pseudogenes in *Drosophila*

Lisa M. J. Bardsley, Wayn Wong, Charles Robin

Department of Genetics, The University of Melbourne, Victoria, 3010 Australia.

Pseudogenes are genomic sequences which have been derived from functional genes but which are themselves no longer functional. They are presumed to evolve neutrally, and thus can be used as tools with which to characterize neutral evolution. Whilst humans are estimated to have around 20,000 pseudogenes, attempts to find pseudogenes in *Drosophila* have proven to be far less successful, with one study estimating their number to be around 109 (Harrison et al., 2003). We have performed an in-depth analysis of these 109 pseudogenes along with the sequences annotated as pseudogenes on Flybase (the database for *Drosophila* genetics and molecular biology). Surprisingly, our results have shown that the majority of these sequences are unlikely to be genuine pseudogenes, instead representing functional genes and transposable elements. Reasons for the erroneous designation of these as pseudogenes include incorrect prediction of splice boundaries, failure to recognize novel gene structures, and the presence of null alleles in the sequenced line.

69. Linkage disequilibrium in natural populations: information statistics provide a powerful test.

William B Sherwin[*] Jurgis Sapijanskas [*¶] & Dhriti Pandya[*§]

[*] *Evolution and Ecology research Group, School of Biological Earth and Environmental Science, University of NSW, Sydney NSW 2052 Australia*

[¶] *Ecole Polytechnique 91128 Palaiseau Cedex Paris France*

[§] *Austin College, Sherman, Texas, USA (current address). Texas Academy of Mathematics and Science, University of North Texas, USA*

Wild studies of gametic (or "linkage") disequilibrium are useful for fine-scale gene mapping, for investigations of multilocus selection, and forensics. However, a common problem in wild populations is that linkage phase is usually unknown, even in many human datasets. Here we compare a number of statistics for summarising and/or testing disequilibrium in wild populations: D, D', r^2, Z, and three new measures from Information theory. We simulated each measures' ability to express association between alleles at different loci when the population was close to, or far from, equilibrium. We evaluated measures on their allele proportion dependency, ability to correctly rank different linkage cases, robustness to random genetic drift, power in statistical tests, and performance with phase-unknown data. Under a variety conditions, r^2 provided the best summary of the disequilibrium. However, in the common case where linkage phase is unknown, an information-based G-statistic was the most powerful statistical test, with low rates of false discovery and false rejection. We demonstrate the utility of the new method using data on haplotypes near the *BRCA2* locus in humans. This new method enhances power of analyses of genotypic disequilibrium in wild populations.

M. J. D. White Address

70. Food, sex, drugs and death in fungi: identification of genes involved in nutrient sensing and programmed cell death

Margaret E. Katz

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 hexokinases (HxkC and HxkD) and the third gene product (XprG) belongs to a newly defined class of p53-like DNA-binding proteins. 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. Preliminary evidence suggests that HxkC may block XprG-mediated PCD. *hxkCΔ* null mutants undergo spontaneous PCD, as shown by fragmentation of DNA and high levels of intracellular protease (a marker of autophagy). Both of these PCD markers are suppressed in *hxkC- xprG-* double mutants. We have used microarrays to investigate which genes are regulated by XprG in response to carbon starvation. The results show that expression of many genes is greatly reduced in an *xprGΔ* null mutant, indicating that XprG may play a major role in the response to carbon limitation. Many of the genes with altered expression (e.g. genes in the aflatoxin biosynthetic pathway) are known to be regulated in response to carbon starvation. Our studies suggest that the regulatory pathway that we have identified controls the expression of fungal virulence determinants and may be a useful target for anti-fungal agents.

Margaret Katz received a Ph.D. in Biological Sciences from the University of California at Santa Barbara for research in microbial genetics. As a post-graduate student she studied genes involved in cell division control and mating in yeast and mating type switching in the ciliate *Tetrahymena thermophila.* She was introduced to the study of gene regulation in *Aspergillus nidulans* (so far, a continuing life-long interest!) as a post-doctoral research fellow in the laboratory of Professor Michael Hynes at the University of Melbourne. During a second post-doctoral fellowship at Monash University in the laboratory of Professor Julian Rood she identified genes which could be used to discriminate between virulent and benign isolates of the bacterial pathogen which causes footrot in sheep. In 1991, she accepted a position as Lecturer in Molecular Genetics at the University of New England in Armidale, NSW. Her research is currently focussed on gene regulation in filamentous fungi, diagnosis of bacterial and fungal diseases, and bacterial and fungal genes involved in virulence. She is currently an Associate Professor at the University of New England.

Tuesday 8 July 2008, Eclipse Room, Union Building

17.00 – 19.00 POSTERS AND COCKTAILS

LIST OF POSTERS (number indicates display board position)

1) Ali, Shahid, Phill Batterham and Trent Perry. *The role of nicotinic acetylcholine receptors and their regulation in spinosad resistance*

2) Arya, Piyush and Priya Chatterjee. *Proteomics in translational cancer research*

3) Chan, Jackie T., Tom J. Dixon, William B. Sherwin, Iain M. Suthers, and Matthew D. Taylor. *Establishing a genetic basis for the responsible stock enhancement of the eastern king prawns (Penaeus Melicertus plebejus)*

4) Chang, Leiyao, Wayne Pitchford and Cynthia Bottema. *Locating and characterising genes affecting beef tenderness*

5) Chatterjee, Priya. *Gene Expression Data Analysis and Modeling*

6) Chong, Josephine YT, Chandramaya Siska Damayanti, Stephen Kolomyjec, Tom Grant, Jaime Gongora and Chris Moran. *Genetic population studies of platypus (Ornithorhynchus anatinus) in South-Eastern New South Wales*

7) Daish, Tasman, Frank Grützner and Enkhjargal Tsend-Ayush. *Meiotic sex chromosome inactivation in monotremes (now moved oral presentation, Abstract 64a) there will be no poster*

8) Ezaz, Tariq, Alexander E. Quinn, Stephen D. Sarre, Arthur Georges, Denis O'Meally, Jennifer A. Marshall Graves. *ZooFISH reveals homologous sex chromosomes among GSD and TSD Australian dragons*

9) Fellfort, Geesa, Embel du Ped, and Avril F. Hoques. *In the beginning was the Word: Canonical language patterns in biomolecules*

10) Gwilliam, Jessica, Adam Stow, Robert Harcourt. *Conservation genetics of commercially exploited sharks*

11) Harvey, Susan. *Phylogeography of the Grassland Melomys (Melomys burtoni)*

12) Honda, Takahiro, Narelle E. Tunstall and Coral G. Warr. *Genetic screens to identify olfactory genes in Drosophila*

13) Hunter, A., Jin, B, and Kelly, JM. *Genetic modification of Aspergillus oryzae for treatment of winery wastes.*

14) Jaratlerdsiri, Weerachai, Chandramaya Siska Damayanti, Lee Miles, Lorna Melville, Chris Moran, Sally Isberg and Jaime Gongora. *Three groups of Endogenous Retroviruses found in Crocodilians*

15) Jitklang, Sanae, Chaliow Kuvangkadilok, Visut Baimai, Hiroyuki Takaoka, and Peter H. Adler. *Cytogenetics and Morphotaxonomy of the Simulium (Gomphostilbia) ceylonicum species group (Diptera: Simuliidae) in Thailand.*

16) Kilinc, Aydin and David R. Smyth. *Evolution of an enhancer region that regulates PETAL LOSS expression in the perianth of the Arabidopsis flower.*

17) Klarić, Thomas, Martin Lewis, Murray Whitelaw and Simon Koblar. *The role of Nxf in neurogenesis*

18) Lawlor, K.T. , C.L. van Eyk, S. Samaraweera, L.V. O'Keefe, C.J. McLeod, S. Dayan, and R.I. Richards. *Pathways of pathogenesis in dominant expanded repeat diseases*

19) Lim, Hiendleder, Fitzsimmons, Grützner. *DNA replication analysis of polyploid bovine trophoblast giant cells*

20) Cintia M. Martinez-Garduno, Yvonne M. Parsons. *Selection experiments under laboratory conditions to ZnCl2 in Chironomus tepperi*

21) Mitchell, Judith, Phil Daborn, Phil Batterham, Trent Perry. *Nicotinic Acetylcholine Receptors and their Roles in Insecticide Resistance*

22) Mohammadi, Amir, Margaret Delbridge, Janine E. Deakin, Paul D. Waters, and Jennifer A. M. Graves. *Characterization of an MHC-linked olfactory receptor gene cluster in Tammar wallaby*

23) Murtagh, Veronica J., Paul D. Waters, Jennifer A. Marshall Graves. *Revealing the wallaby Y chromosome*

24) Myers, Steven. *Comparing Morphology and Gene flow in New Holland Honeyeaters at a Cross-regional scale*

25) Navdeep, Bhatti. *Chromosome and genome size evolution in the genus Schoenus*

26) Novianti, Irida, Wayne Pitchford, and Cynthia Bottema. *Molecular genetics of cattle body conformation*

27) O'Keefe, Louise, Alex Colella, Sonia Dayan, Qingwen Chen and Robert Richards. *Common fragile site-associated genes in Drosophila*

28) Pornpimol P-Rattanatrai, Chandramaya Siska Damayanti, Chris Moran, Jaime Gongora. *Emergence of MHC non-classical class Ib genes in pigs and peccaries*

29) Potter, Sally David Taggart, Steve Cooper and Mark Eldridge. *Phylogeography and population genetics of rock-wallabies in the Kimberley, WA*

30) Stark, Alan E. *Mating and offspring frequencies under partial outcrossing in a structured population*

31) Swan, Amelia, Chandramaya Siska Damayanti, Stephen Kolomyjec, Tom Grant, Chris Moran and Jaime Gongora. *Mitochondrial DNA analysis of Platypus from New South Wales*

32) Tangkawanit, Ubon, Chaliow Kuvangkadilok , Visut Baimai and Peter H Adler. *Cytogenetics of the Simulium tuberosum group (Diptera: Simuliidae) in Thailand.*

33) Zarnegar, Armita, Mohammed M Islam, Peter Vamplew, Andrew Stranieri. *A hybrid evolutionary algorithm for nucleosome positioning*

34) Zulkifli, Nadiatur Akmar, Madan Naik, Wayne Pitchford and Cynthia Bottema. *Molecular Genetics of Mitochondrial Function and Net Feed Efficiency in Cattle*

35) William G. Alexander, Namboori B. Raju, Hua Xiao, Thomas M. Hammond, Tony D. Perdue, Edward G Barry, Robert L. Metzenberg, Jason E. Stajich, Patricia J. Pukkila, and Patrick K.T. Shiu. *Relationships between chromosome pairing and meiotic silencing of unpaired DNA (MSUD) in Neurospora crassa*

36) Naomi Ratcliff, Chandramaya Siska Damayanti, Chris Moran, Sally Isberg, Jaime Gongora. *Mitochondrial control region analyses of the Saltwater Crocodile from the Northern Territory.*

37) Elizabeth P. Murchison[1*], Pouya Kheradpour[2], Ravi Sachidanandam[1], Carly Smith[1], Zhenyu Xuan[1], Frank Grützner[4], Alexander Stark[2,3], and Gregory J. Hannon[1] MiRNAs and piRNAs in platypus and echidna.

ABSTRACTS OF POSTERS

Poster 1. THE ROLE OF NICOTINIC ACETYLCHOLINE RECEPTORS AND THEIR REGULATION IN SPINOSAD RESISTANCE

Shahid Ali, Phill Batterham and Trent Perry

Genetics Department, Bio-21 institute, University of Melbourne, Victoria 3010

Resistance to insecticides conferred through target modification is a serious problem important agricultural pest species of the world. The Spinosad insecticide is a fermentation product of *Saccharopolyspora spinosa*. Due to its extensive use, resistance to spinosad has been reported by House fly, Tobacco budworm, Diamond back moth, Beet armyworm and Fruit fly. Spinosad targets the nicotinic acetylcholine receptors (nAChR) and GABA-gated chloride channels. It allosterically activates nAChR and prolongs the acetylcholine responses and it inhibits the normal function of the GABA receptors. My project aim is to characterize the expression of nAChR subunit genes in Drosophila. Constructs containing 5' regions of Da3, Da4, Da5, Da7, Db1 and Db3 subunits will be created. These will be crossed with UAS:GFP strains and the expression of these subunits will be analysed in conjunction with RNA insitus. The studies will determine when receptor subunits will be available as a target for insecticides such as spinosad and if these could potentially participate in the same receptor. Knock down will be carried out for these receptors using RNAi lines. If these found to be resistant to spinosad then EMS mutagenesis will be carried out to generate resistance alleles.

Poster 2. PROTEOMICS IN TRANSLATIONAL CANCER RESEARCH

Piyush Arya and Priya Chatterjee

Biotechnology, Vellore institute of technology, D-324, Vellore, Tamil nadu, India, 632014

The sequencing of the human and other important genomes is only the beginning of the quest to understand the functionality of cells, tissues, and organs, both in health and disease. Together with advances in bioinformatics, this development has paved the way to the revolution in biology and medicine that we are experiencing today. We are rapidly moving from the study of single molecules to the analysis of complex biological systems, and the current explosion of emerging technologies within proteomics and functional genomics promises to elicit major advances in medicine in the near future. Cancer, being a complex disease that affects a significant fraction of the population, is foreseen as a prime target for the new technologies, as tools for the high throughput analysis of genes and proteins might expedite the applications of basic research findings into daily clinical practice through translational research. In particular, proteomic technologies are expected to play a key role in the study and treatment of cancer, as they provide invaluable resources to define and characterize regulatory and functional networks, to investigate the precise molecular defect in diseased tissues and biological fluids, and for developing specific reagents to precisely pinpoint a particular disease or stage of a disease. For drug discovery, proteomics assist with powerful tools for identifying new clinically relevant drug targets, and provide functional insight for drug development.

Proteomics encompasses many platform technologies for protein separation and identification, for determining their biomolecular interactions, function, and regulation, and for annotating, storing, and distributing protein information. The proteome is much more complex and dynamic than the genome, and the task of deciphering it even in a single cell type is daunting, as there may be many thousands of proteins, including splice variants, posttranslational modifications, and cleavage products, present in a cell at any given time. Moreover, the dynamic range of protein expression expands over several orders of magnitude, a fact that limits their characterization and analysis.

In this paper we present proteomics as a platform technology for analysis of tissue biopsies.

Poster 3. Establishing a genetic basis for the responsible stock enhancement of the eastern king prawns (*Penaeus Melicertus plebejus*)

Jackie T. Chan[1*], Tom J. Dixon[2], William B. Sherwin[1], Iain M. Suthers[1,3], and Matthew D. Taylor[1,3]

[1]Ecology and Evolution Research Centre, School of Biological, Earth and Environmental Science, University of New South Wales, Sydney 2052 Australia. [2]Food Futures Flagship, CSIRO Livestock Industries, Qld Bioscience Precinct, 306 Carmody Rd, St Lucia, QLD 4067, Australia. [3]Sydney Institute of Marine Science, Building 22, Chowder Bay Road, Mosman, NSW, 2088, Australia. [*]Corresponding author: jackie.t.chan@gmail.com

Eastern king prawn (EKP) is an important commercial prawn species in Australia, distributed

from northern Queensland to southern New South Wales. Assessment of the status and structure of this species may provide a basis for an ongoing stock enhancement program to increase and maintain population size of EKP in recruitment limited estuaries. Evaluation of the genetic impacts of releasing hatchery-reared post-larvae is essential for responsible large scale stock enhancement. This project aims to develop a genetic system for assessing the current stock structure, identifying the origin of recaptured prawns, and detecting genetic impacts of stock enhancement. The project will commence with development of suitable genetic markers, and then apply these markers to address the above objectives. The potential genetic concerns for stock enhancement programs in general, and the possible impact on the natural EKP population are considered.

Poster 4. Locating and characterising genes affecting beef tenderness

Leiyao Chang[1], Wayne Pitchford[1,2], and Cynthia Bottema[1,2]

[1]Discipline of Agriculture and Animal Science, The University of Adelaide, Roseworthy, SA, 5371.[2]CRC for Beef Genetic Technologies

Tenderness is regarded as one of the most important palatability attributes of beef and a major sensory factor that affects re-purchase intent of meat by consumers. Six candidate genes, which were selected based on QTL analysis results, have been investigated for their effect on tenderness. One gene, that is believed to be involved in cross-linking within the extracellular matrix of muscles, appears to significantly affect the tenderness of the *longissimus* muscle (LD). Moreover, another gene, which is a protease, has been discovered to significantly influence the tenderness of semitendinosus muscles (ST). Although both of these genes affect tenderness, there is no interaction between them. Further studies are underway to investigate additional genes that contribute to tenderness and to test for interactions between the genes in order to identify physiological or molecular pathways determining beef tenderness.

Poster 5. Gene Expression Data Analysis and Modeling

Priya Chatterjee

Biotechnology, Vellore institute of technology, D-324, Vellore, Tamil nadu, India, 632014

The traditional approach to research in Molecular Biology has been an inherently local one, examining and collecting data on a single gene, a single protein or a single reaction at a time. However, with the advent of the "Age of Genomics" an entirely new class of data is emerging. can we really expect to construct a detailed biochemical model of, say, an entire yeast cell with some6000 genes (only about 1000 of which were defined before sequencing started, and about 50% of which are clearly related to other known genes), by analyzing each gene and determining all the binding and reaction constants one by one? Likewise, from the perspective of drug target identification for human disease, we cannot realistically hope to characterize all the relevant molecular interactions one-by-one as a requirement for building a predictive disease model.

There is a need for methods that can handle this data in a global fashion, and that can analyze such large systems at some intermediate level, without going all the way down to the exact biochemical reactions. Gene sequence information in cis regions (regulatory inputs) and protein coding regions (regulatory outputs; determines biomolecular dynamics) is expanded into spatio-temporal structures defining the organism. Some principles of this behavior may be captured by computational models, such as Boolean networks. In order to draw meaningful inferences from gene expression data, it is important that each gene is surveyed under several different conditions, preferably in the form of expression time series. Such data sets may be analyzed using a range of methods with increasing depth of inference, such as cluster analysis, correlation analysis, and determination of mutual information content.

Poster 6. Genetic population studies of platypus (*Ornithorhynchus anatinus*) in South-Eastern New South Wales

Josephine YT Chong[1], Chandramaya Siska Damayanti[1], Stephen Kolomyjec[2], Tom Grant[3], Jaime Gongora[1] and Chris Moran[1]

[1]Centre for Advanced Technologies in Animal Genetics and Reproduction, Faculty of Veterinary Science, RMC Gunn Building B19. University of Sydney, NSW 2006, Australia. [2]School of Marine and Tropical Biology, James Cook University, Townsville, QLD, Australia. [3]School of Biological, Earth and Environmental Sciences, University of New South Wales, Australia 2052.

Endemic to Australia, the platypus (*Ornithorhynchus anatinus*) is one of three surviving monotreme

species and is ecologically and genetically interesting. Being semi-aquatic, the species is restricted to permanent freshwater rivers ranging from eastern Queensland and New South Wales to eastern, central and south-western Victoria, King Island and Tasmania. The disjunct and broad distribution of the platypus in rivers may have led to population structure. However, there is limited knowledge of platypus population genetics. This study will present preliminary results of microsatellite analyses from platypus from four locations in southern NSW. We are currently in the process of optimising ten microsatellite markers to be used for genotyping tagged individuals. This study will provide genetic information on population structure and allele diversity to assist in conservation and management plans for the platypus and help ensure its survival.

Poster 7. changed to oral presentation

Poster 8. ZooFISH reveals homologous sex chromosomes among GSD and TSD Australian dragons

Tariq Ezaz[1,2], Alexander E. Quinn[2], Stephen D. Sarre[2], Arthur Georges[2], Denis O'Meally[1], Jennifer A. Marshall Graves[1]

[1]Comparative Genomics Group, Research School of Biological Sciences, Australian National University. [2]Institute for Applied Ecology, University of Canberra

Sex in Australian dragon lizards (Agamidae) is determined by either genes (Genetic Sex Determination, GSD) or by the influence of temperature on the developing embryo (Temperature-dependent Sex Determination, TSD) – and in some species both interact to determine sex. The close evolutionary relationship of Australian dragon lizards and their rapid radiation makes them an ideal group in which to study transitions between GSD and TSD. Indeed, some sister species show alternate modes of sex determination, suggesting frequent transitions. Karyotypes are highly conserved. No sex chromosomes can be distinguished cytologically in GSD species, but differential staining techniques reveal Z and W microchromosomes. Recently, we identified a novel sex-specific 3.3kb genomic sequence from the W chromosome of the central bearded dragon, *Pogona vitticeps*, by AFLP and subsequent genome walking procedures. We screened both GSD and TSD dragons using this sequence as a FISH probe. Hybridisation on a single pair of chromosomes in each species examined indicates that sex chromosome sequences are conserved, implying homologous chromosomes between GSD and TSD dragons. Our findings revealed that the same microchromosome pair is retained in GSD and TSD dragons, and has become sex specific in GSD species.

Poster 9. In the beginning was the Word: Canonical language patterns in biomolecules

Geesa Fellfort, Embel du Ped, and Avril F. Hoques

Center for Science and Culture, Discovery Institute, 208 Garden Path, Seattle, WA 98104, USA.

English language words sometimes occur in the sequences of proteins written in the one-letter amino acid code. In some synthetic proteins or protein domains, this is because the sequence carries instructions such as for protein import to an organelle [1]. In natural proteins, the frequencies at which such words are found varies considerably depending on the word. For instance, the pentapeptide NH2-Glu-Leu-Val-Ile-Ser-COOH (ELVIS) is at least four times more frequent in the protein sequence database than its isomer LIVES and it is not self-evident whether natural selection can fully account for this observation [2, 3]. The capacity of DNA and RNA sequences to carry information is not limited to the proteins they encode. It is possible that a gene could code for meaningful text in unused reading frames. This property may have been used by gene designers to place watermarks [4, 5], messages [1, 6, 7], or other canonical lexical content in sequences. To test this hypothesis, and to characterize the lexicomes of different species, we developed the Bolex algorithm. We found a statistically significant excess of English language *k*-mers in DNA and mRNA sequences from some avian taxa. The most striking of these is a *Geospiza fortis* mRNA [8] that contains out-of-frame matches to 65 different English words of up to 11 letters long (there are two instances of the string MAIDSERVANT), as well as many near-words such as ADVLTERY. Since the origin of the English language can be traced to less than 6000 years ago, we are currently developing methods to map biomolecular sequences onto languages with non-Roman alphabets such as Aramaic and Hebrew.

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4.Marillonnet S *et al.* (2003). Nat Biotechnol *21*, 224.
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8.Moses A (2008). GenBank accession number AM950316.

Poster 10. Conservation genetics of commercially exploited sharks

Jessica Gwilliam[1], Adam Stow[1], Robert Harcourt[2]

[1]*Department of Biological Sciences, Macquarie University, Sydney 2109.*[2]*GSE, Macquarie University, Sydney 2109*

The proposition that low genetic variation increases the risk of disease has had little consideration in the marine environment. Commercial exploitation has lead to declines in shark numbers worldwide. While improved management has seen partial recovery of previously overfished species, such as gummy, *Mustelus antarcticus* and school shark, *Galeorhinus galeus,* it is unclear how past overexploitation has affected the genetic diversity and health of these populations. In this study we assess genetic structure and diversity in gummy and school shark. We will first characterize the population structure of these two species and establish whether there are differences in genetic variation among regions. *A priori* we expect that low genetic variation will be present in some localities where populations have suffered significant declines. The relationship between disease and genetic variation will be investigated by collecting data on the presence and abundance of particular pathogens such as myxosporean parasites. Finally, mating strategies will be determined as these influence effective population size, and therefore the rate at which we might expect genetic variation to be eroded.

Poster 11. Phylogeography of the Grassland Melomys (Melomys burtoni)

Susan Harvey

Natural Resource Sciences, Queensland University of Technology, Queensland, Australia

Two mitochondrial and one nuclear DNA markers were targeted to investigate the phylogeographic history of *Melomys burtoni*. Genetic structure was overlaid on geographical information in order to tease apart the contemporary and historical processes that have shaped the current distribution. Results have revealed that several factors have had an important influence on the phylogeography of *M. burtoni*. Pleistocene sea level fluctuations allowed gene flow between Australia and New Guinea via a land bridge either side of Lake Carpentaria. Four regions have been identified where zoogeographic barriers have restricted gene flow among *M. burtoni* populations. These include the Wet Tropics region, the Kimberley-Arnhem Land barrier, another unidentified zoogeographic barrier between populations in western and eastern Northern Territory and the Gulf Plains region around the coastline of the Gulf of Carpentaria. As a result of the Gulf Plains barrier, as *M. burtoni* expanded from New Guinea into Australia either side of Lake Carpentaria there was no secondary mixing at the base of the Lake, with gene flow between east and west Australia occurring via New Guinea. *M. burtoni* is likely to have an extralimital distribution in New Guinea as *M. lutillus* and a formal taxonomic revision including a morphological assessment is required.

Poster 12. GENETIC SCREENS TO IDENTIFY OLFACTORY GENES IN *DROSOPHILA*

Takahiro Honda, Narelle E. Tunstall and Coral G. Warr

School of Biological Sciences, Monash University, Australia

In *Drosophila* odour signals are detected in olfactory receptor neurons (ORNs) by a large family of 62 seven-transmembrane receptor proteins, the odorant receptor (Or) family. Initially Ors were thought to be G protein-coupled receptors, however recent studies indicate they have a different membrane topology.

Poster 13. Genetic modification of *Aspergillus oryzae* for treatment of winery wastes.

Hunter, A[1,2], Jin, B[2], and Kelly, JM[1]*

[1]*School of Molecular and Biomedical Science, The University of Adelaide, South Australia, 5005 Australia.* [2]*School of Earth and Environmental Science, The University of Adelaide, South Australia, 5005 Australia.*

The filamentous fungus *Aspergillus oryzae* has been used in Oriental food production for centuries, and its ability to secrete high levels of endogenous or heterologous enzymes facilitates its extensive use in the fermentation industry. We aim to utilise *A. oryzae* in a treatment process to convert glucose-

rich winery wastes to protein-rich fungal biomass for use as livestock feed. In the model fungus *Aspergillus nidulans*, disruption of *creB* encoding a deubiquitinating enzyme leads to a relaxation of glucose repression, and elevated expression of a range of enzymes for the utilisation of a range of carbon sources. Here we present the deletion of *creB* in two strains of *A. oryzae*: RIB40, the strain that had its genome sequenced, and DAR3699, a strain with morphological properties well-suited to fermentation. We analyse the effects of *creB* deletion on the secretion of amylases, cellulases and other enzymes both in the presence and absence of glucose, and discuss implications for the use of *A. oryzae* in the bioconversion of wastes.

Poster 14. Three groups of Endogenous Retroviruses found in Crocodilians

Weerachai Jaratlerdsiri[1], Chandramaya Siska Damayanti[1], Lee Miles[1], Lorna Melville[2], Chris Moran[1], Sally Isberg[3] and Jaime Gongora[1]*.

*[1]Centre for Advanced Technologies in Animal Genetics and Reproduction, Faculty of Veterinary Science, RMC Gunn Building B19. University of Sydney, NSW 2006, Australia. [2]OIC Berrimah Veterinary Laboratories, Department of Primary Industry, Fisheries and Mines, GPO Box 3000 Darwin 0801. [3]Porosus Pty Ltd, PO Box 86, Palmerston NT 0831, Australia. *Corresponding author: j.gongora@usyd.edu.au*

Endogenous retroviruses (ERVs) are copies or remnants of exogenous retroviruses that were integrated into the host genome at some stage in the past. ERVs are vertically transmitted from the host to its progeny. Although most ERVs are defective due to inactivating mutations, functional ERVs are potential agents of disease. Previous studies have identified the presence of a distinct clade of ERV from six species of crocodilians. Here we analyse the functionality, distribution and phylogenetic relationships of ERVs in twenty extant species of crocodilians (crocodiles and alligators) from across the world. The ERV reverse transcriptase (*pol*) gene fragment (0.8-1 kb) was amplified, cloned and sequenced. Preliminary analyses of sixty-six crocodilian ERV *pol* DNA sequences show that these retroelements possess stop codons and deleterious mutations. Thus, crocodilian ERVs are generally, if not universally, defective as has been observed in many other hosts. Phylogenetic analyses show that crocodilian ERVs cluster in three major clades, one crocodile-specific and one alligator-specific. Surprisingly, ERV sequences from *Crocodylus palustris* cluster within a third clade consisting mainly of alligator sequences. Further analyses of the evolutionary relationships and evidence for ERV functionality within crocodilians are underway.

Poster 15. Cytogenetics and Morphotaxonomy of the *Simulium* (*Gomphostilbia*) *ceylonicum* species group (Diptera: Simuliidae) in Thailand.

Sanae Jitklang*, Chaliow Kuvangkadilok, Visut Baimai, Hiroyuki Takaoka, and Peter H. Adler

Department of Biology, Faculty of Science, Mahidol University, Bangkok 10400, Thailand.

The polytene chromosomes of 1,612 larvae of three described morphospecies in the *Simulium* (*Gomphostilbia*) *ceylonicum* species group – *S. asakoae*, *S. inthanonense*, and *S. sheilae* – were examined from 52 sites in Thailand. A standard map for the *S. ceylonicum* group was established. Ten cytoforms, plus an eleventh from the literature, are revealed on the basis of unique suites of fixed and floating inversions; sex chromosomes are microscopically undifferentiated in all cytoforms. A cytodendrogram, based on shared inversions, shows seven lineages in a polytomy derived from a hypothetical ancestor. Morphological descriptions of the known life stages of each cytoform and keys to larvae, pupae, and polytene chromosomes also are provided. Despite small sample sizes for some cytoforms, all segregates appear to be good species, supported by both chromosomal and morphological evidence. Three reproductively isolated cytoforms for which adequate material is available are formally described as new species. The existence of chromosomally distinct entities within established morphospecies of the *S. ceylonicum* group supports a recurrent trend of hidden biodiversity in Southeast Asian black flies. The greater diversity of differentiated taxa that we found in the *S. ceylonicum* group in northern, versus southern, Thailand, along with differences in taxa between northern and southern Thailand, conforms to a similar biogeographic trend in diverse organisms.

Poster 16. Evolution of an enhancer region that regulates *PETAL LOSS* expression in the perianth of the *Arabidopsis* flower

Aydin Kilinc and David R. Smyth

School of Biological Sciences, Monash University, Victoria 3800, Australia.

An outstanding issue in biology is the evolution of complex forms, and how changes in regulatory

networks drive this process. Flower structure in the Brassicaceae is remarkably conserved. The outermost whorl consists of four sepals, and the second whorl contains four petals arising between them, giving a distinctive cross-shaped appearance.

The *PETAL LOSS* (*PTL*) gene encodes a transcription factor of the trihelix family with 29 members in *Arabidopsis*. The *ptl* mutant phenotype is characterized by aberrant sepal development and a partial loss of petals. *PTL* is expressed between developing sepal primordia (termed the inter sepal zone, ISZ) where it suppresses growth. This keeps the sepals separate, and also limits the size of nearby sites for petal initiation.

We have identified a highly conserved enhancer region within the single long intron of *PTL*, and have shown that it activates expression in the ISZ. However, this sequence is apparently absent in the intron of PTL orthologs from outside the order Brassicales. Here we show that, within the Brassicales, a switch from 5-fold to 4-fold symmetry of the perianth coincides with the presence of a long intron containing the enhancer region. Furthermore, this region shows close homology to a sequence within the LTRs of a *gypsy-like* retrotransposon family, and that simple mutation in the LTR sequence is capable of activating expression in the ISZ. PTL function may have been recruited to the ISZ regions of the flower via the insertion of a retroelement into the intron of an ancestral *PTL* gene, and that this was associated with a change from pentamery to tetramery.

Poster 17. The role of *Nxf* in neurogenesis

Thomas Klarić, Martin Lewis, Murray Whitelaw and Simon Koblar

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

Nxf, also called *LE-PAS* or *NPAS4*, is a brain specific transcription factor of the basic helix-loop-helix PAS family. Members of this family are typically involved in developmental processes and cellular response to environmental stresses. *Nxf* is expressed in neurogenic regions of the adult brain including the hippocampus and olfactory bulb and is up-regulated in response to various stresses such as seizure, cerebral ischaemia and cortical spreading depression. Animal models of brain injury have shown that both seizure and cerebral ischaemia lead to increased neural stem cell proliferation and neurogenesis. Similarly, cortical spreading depression can also stimulate neurogenesis. Most recently, it was shown that mice raised in social isolation had reduced levels of *Nxf* expression. These mice had decreased survival of newly differentiated neurons in the hippocampus and impaired learning. Taken together these data suggest a hypothesis that *Nxf* may transcriptionally regulate neurogenesis in the adult mammalian brain following environmentally induced stress. We have used neuronal differentiation of mouse embryonic stem cells as an *in vitro* model of neurogenesis to investigate the role of *Nxf* in this process. We have characterised the expression profile of *Nxf* over the course of differentiation and observed an increase in *Nxf* expression associated with differentiation.

Poster 18. Pathways of pathogenesis in the dominant expanded repeat diseases

K.T. Lawlor[1], C.L. van Eyk[1], S. Samaraweera[1], L.V. O'Keefe[1], C.J. McLeod[1], S. Dayan[1], and R.I. Richards[1,2].

[1]ARC Special Research Centre for the Molecular Genetics of Development, [2]ARC-NHMRC Research Network in Genes and Environment in Development, School of Molecular and Biomedical Sciences, The University of Adelaide, S.A. 5005, Australia.

Dominant expanded repeat diseases are caused by the expansion of a simple nucleotide repeat beyond a pathogenic threshold. They include neurodegenerative diseases caused by the expansion of a polyglutamine coding CAG repeat such as Huntington's, SBMA and Spinocerebellar Ataxias (SCA) 1,2,3,6,7 and 17. Expanded repeat sequences within non-coding regions can also give rise to neurodegenerative disease. Examples include SCA 8,10 and 12, Huntington's disease-like 2, Myotonic Dystrophy Type 1 and 2 (DM1 and DM2) and Fragile X Tremor Ataxia Syndrome. Since the untranslated and translated dominant expanded repeat diseases seem to share a number of clinical features and have a shared mutation mechanism we hypothesise that there may be common pathogenic pathways between the two disease classes. We are modelling a number of these diseases using *Drosophila* to identify the molecular basis of pathogenesis and the contribution of hairpin RNA structure.

McLeod CJ, O'Keefe LV, RI Richards (2005) The pathogenic agent in *Drosophila* models of polyglutamine diseases. *Hum Mol Genet., 14(8): 1041-8.*

Poster 19. DNA replication analysis of polyploid bovine trophoblast giant cells

Lim, Hiendleder, Fitzsimmons, Grützner

The University of Adelaide, 5005, Adelaide, South Australia

Trophoblast giant cells (TGCs) play an important role in mammalian placentas. They synthesise and secret hormones important for foetal development and adaptation to pregnancy. Genome multiplication is a typical feature of TGC development in mammals. In bovine TGCs genome multiplication was found in placentomal and interplacentomal cells but is largely missing in the maternal uterine epithelium cells of the placentome. Fluorescence In situ hybridisation with several autosomal BAC clones was used to investigate the ploidy level of cultured TGCs. This shows that 84% of the cultured TGC are tetraploid and 6% of cells are either octaploid or feature even higher ploidy levels. Genome multiplication increases the copy number, which may also result in overall increased transcript levels. An interphase dot assay (IDA) was performed to investigate if a change in replication timing occurs in TGCs. This revealed that in almost half of the tetraploid TGCs tested the replication pattern changes from synchronous replication in fibroblast cells to asynchronous replication pattern in TGCs. This indicates that silencing of additional copies of genes may occur in polyploid TGCs.

Poster 20. Selection experiments under laboratory conditions to ZnCl2 in Chironomus tepperi

Cintia M. Martinez-Garduno, Yvonne M. Parsons

Department of Genetics and Human Variation, La Trobe University, Victoria, 3086, Australia

Selection experiments are an essential tool to understand the evolvability of specific traits and are particularly important in ecotoxicological approaches to study the response of key species towards toxicants over several generations.

We are interested in understanding the tolerance of chironomid species to heavy metals, as it has been shown that species-specific variation exists.

We are conducting selection experiments in Chironomus tepperi, exposing larvae to ZnCl2 under laboratory conditions. Although the actual genes involve in zinc tolerance are unknown, previous studies have suggested a genetic response to selection in the populations.

On the first generations analyzed, we have observed that selection experiments have shown significant frequency difference in 14/115 AFLP loci. However, there is also the possibility that the genetic changes are due to other effects, such as inbreeding rather than the selection regime. To determine the actual cause of these changes, analyses of allelic frequency and genetic variation on each generation are being conducted.

Poster 21. Nicotinic Acetylcholine Receptors and their Roles in Insecticide Resistance

Judith Mitchell, Phil Daborn, Phil Batterham, Trent Perry

Centre for Environmental Stress and Adaptation Research, Genetics Department, University of Melbourne, Victoria, Australia.

The nicotinic acetylcholine receptor family in *Drosophila melanogaster* includes ten genes which are expressed in different combinations in order to produce mature receptors. These receptors have been found to be effective targets for two distinct insecticide classes, the neonicotinoids and the spinosyns. Specifically, neonicotinoids appear to bind to the subunits Da1 and Db2 (PERRY *et al.* 2008), while the spinosyns target Da6 (PERRY *et al.* 2007). Resistance to either of these insecticides can occur when one of the subunits it binds to is knocked-out. Loss of any one of these three receptor subunits does not appear to affect viability of the insect. We are using targeted RNAi knock-down to examine whether any of the nAChR subunits are essential for viability in *D. melanogaster*. The impact of altered expression of subunits on the response seen to insecticides is being examined, and may uncover additional subunits that are involved in insecticide binding.

PERRY, T., D. G. HECKEL;, J. A. MCKENZIE; and P. BATTERHAM, 2008 Mutations in Da1 or Db2 nicotinic acetylcholine receptor subunits can confer resistance to neonicotinoids in Drosophila melanogaster. Insect Biochem Molecular Biology.

PERRY, T., J. A. MCKENZIE and P. BATTERHAM, 2007 A Da6 knockout strain of Drosophila melanogaster confers a high level of resistance to spinosad. Insect Biochemistry and Molecular Biology **37:** 184-188.

Poster 22. Characterization of an MHC-linked olfactory receptor gene cluster in Tammar wallaby

Amir Mohammadi*, Margaret Delbridge, Janine E. Deakin, Paul D. Waters, and Jennifer A. M. Graves

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

Olfactory receptor (OR) loci frequently cluster and are present on most chromosomes. They are members of the seven transmembrane receptor (7-TM) superfamily and, as such, are part of one of the largest mammalian multigene families, with an estimated copy number of up to 1000 ORs per haploid genome. As their name implies, ORs are known to be involved in the perception of odors and possibly also in other functions not related to olfaction. One of the OR gene clusters is linked to MHC class I genes in almost all species. This cluster is of particular interest because of its possible involvement in olfaction-driven mate selection. MHC class I genes in most species studied to date are grouped together except for tammar wallaby are split over several chromosomes. It may be that the MHC-Linked OR genes also be located on a number of chromosomes. The major MHC loci in tammar is located on chromosome 2q and some of the genes flanking the OR cluster in opossum have already being mapped to this position. The aim of the study is to map the MHC-Linked OR cluster in tammar wallaby and construct a BAC contig in order to characterize this cluster in marsupials.

Poster 23. Revealing the wallaby Y chromosome

Veronica J. Murtagh, Paul D. Waters, Jennifer A. Marshall Graves

Comparative Genomics Group, Research School of Biological Sciences, The Australian National University, Canberra, ACT, Australia

The mysterious nature of the Y chromosome with its restricted recombination and reduced, yet highly specialized, function is particularly fascinating. The marsupial Y chromosome represents the ancestral mammalian Y, present before the addition of a large autosomal region to the eutherian X and Y. This minimal mammalian Y provides the opportunity for complete characterization that is difficult for the Y chromosomes of humans and other eutherian mammals. The recent publication of the opossum genome, and the completed 2X sequence of the tammar wallaby genome, has provided a vast array of marsupial sequence data that now enables a closer study of the Y chromosome. A tammar wallaby (*Macropus eugenii*) Y chromosome probe was generated by chromosome microdissection and degenerate oligonucleotide polymerase chain reaction (DOP-PCR). This Y probe was used to screen a male wallaby BAC library, resulting in the isolation of 26 BACs that map to the Y chromosome: 16 Y-specific, 8 that map to Y and Xp, 1 that maps to Y and 4p, and a BAC that maps to Y, Xp and 2q. FISH signal variation within this collection of BACs has already begun to give an insight into the repetitive nature of the Y, opening the way for exploration of its gene content and structure. This work will uncover more of the mysteries surrounding the tammar wallaby Y chromosome.

Poster 24. Comparing Morphology and Gene flow in New Holland Honeyeaters at a Cross-regional scale

Steven Myers

Flinders University, South Australia

Conservation strategies are becoming increasingly important with increased concern over the impending issue of global warming and the ever-present effect of anthropogenic activities on the environment. Consequently, the subspecies concept has become increasingly important to identify conservation management priorities (Zink 2004). When ornithologists actively adopted the subspecies classification in the mid-twentieth century, the number of bird species dropped by 50% (Mayr 1970, discussed in Zink 2004). Such a drastic change in taxonomic classification can have significant implications for conservation funding decisions that are often based on species status.

In Australia, honeyeaters (Meliphagidae) are the most diverse and widely distributed of any avian family (Ford *et al.*, 1979; Clarke and Clarke, 1999). They play a key role in the pollination of many endemic plant groups including *Banksia*, *Dryandra*, *Hakea*, *Melaleuca*, and *Eucalyptus* (Driskell and Christidis, 2004). In South Australia, the New Holland Honeyeater (*Phylidonyris novaehollandiae*, NHH) plays a primary role in sustaining the regional ecology. Hence, the loss of these birds heralds the decline of intact ecosystems (Paton *et al.*, 2004), emphasising their importance as a key conservation taxon.

In 1906, Campbell noted a bill length variation, among other morphological differences, between the Kangaroo Island form of NHH and a mainland

form. Subsequently, the KI form (*P. n. Campbelli*) and mainland form (*P. n. novaehollandiae*) are now considered subspecies. However, to date, the state of gene flow between populations in these regions remains to be examined. This project will examine the level of genetic distinctiveness and connectivity between New Holland Honeyeaters from different regions in South Australia. The strength of evolutionary forces on key traits, such as bill morphology, will also be examined.

Poster 25. Chromosome and genome size evolution in the genus *Schoenus*

Bhatti Navdeep

School of Biological Sciences,University of Auckland, Thomas Building, Symond Street, Auckland, New Zealand

Most plants and animals have monocentric chromosomes but large groups amongst plants, such as the Cyperaceae and Juncaceae and animals, the Nematoda and Lepidoptera, have holocentric chromosomes. Holocentric chromosomes have centromeric activity distributed in a diffuse fashion along their entire length. Thus, chromosome breakage has profoundly different consequences for monocentric and holocentric chromosomes. A major question is whether chromosome fusion/fission or polyploidy is responsible for the variation in chromosome number seen in these groups. *Schoenus* contains approximately 100 species, 8 of which are native to New Zealand. These species have chromosome numbers that range from $2n = 8$ to $2n = 74$ with a large variation in chromosome length. Phylogenetic trees based on sequence variation in the internal transcribed region of the 45S rDNA locus were constructed using maximum parsimony and maximum likelihood to reveal evolutionary relationships between species within the genus. Two major clades could be distinguished. *Schoenus* species with large numbers of very small sized chromosomes were grouped together and were found to be ancestral to the other group, comprised of species with small numbers of large chromosomes. Flow cytometry using propidium iodide stained nuclei was used to measure the DNA C-value of different species of the genus. A 12-fold variation in 2C DNA content was found, values ranged from 1.54pg to 18.64pg. There was no obvious relationship between C-value and chromosome number, but a general trend towards genome size increase has been observed. Differential banding with Giemsa and base-specific fluorochromes show a wide variety of banding patterns. Meiosis was regular with bivalent formation in all species examined. Results suggest that chromosome fragmentation has not been involved in the evolution of *Schoenus* genomes.

Poster 26. Molecular genetics of cattle body conformation

Irida Novianti, Wayne Pitchford, and Cynthia Bottema

School of Agriculture, Food and Wine Science, The University of Adelaide, South Australia, 5371 Australia

Body conformation is a phenotypic trait that is widely utilised by farmers to select cattle for high productivity. Body conformation assessment can be accomplished by scoring and measuring traits for either body dimension or muscularity of cattle (e.g. height, length, girth, stifle width and hip width) that are related to muscle development. The objective of this project is to locate the Quantitative Trait Loci (QTL) for body dimension traits in order to identify the chromosomal regions that may contain genes that affect body conformation, and hence, carcass traits. One gene known have a significant role in muscle development is myostatin. Linkage analysis identified 2 QTL controlling body conformation. Interestingly, when myostatin genotype (F94L genotype) was included as an additional fixed effect, one of the QTL was no longer significant. This result suggests that are gene(s) that have epistatic effects with myostatin and these are being investigated for their effect on cattle conformation.

Poster 27. Common fragile site-associated genes in *Drosophila*

Louise O'Keefe, Alex Colella, Sonia Dayan, Qingwen Chen and Robert Richards

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

Common fragile sites are specific regions of chromosomes that are susceptible to breakage after exposure to chemicals in vitro and correspond precisely to genomic rearrangements that have been observed in various cancer patients. Such rearrangements have been shown to result in changed expression levels of the genes found at these sites and altered expression of these fragile site associated genes has been proposed to play a causal role in cancer cell biology. We are using *Drosophila* to determine the normal function of

these fragile site associated genes, many of which are well conserved in this model organism. We have employed homologous recombination to precisely knock out the *FRA16D* associated *Wwox* gene. The resultant *Wwox* mutant flies are viable and fertile and we are undertaking microarray and proteomic analyses to identify putative interactors. We are also looking at other fragile site associated genes (*NitFHIT, Disabled* and *Parkin)* to determine the effect of decreased activity of these fragile site associated genes individually and in combination with each other.

Poster 28. Emergence of MHC non-classical class Ib genes in pigs and peccaries

Pornpimol P-Rattanatrai, Chandramaya Siska Damayanti, Chris Moran, Jaime Gongora*.

*Centre for Advanced Technologies in Animal Genetics and Reproduction, Faculty of Veterinary Science, RMC Gunn Building B19. University of Sydney, NSW 2006, Australia. *Corresponding author: j.gongora@usyd.edu.au*

The major histocompatibility complex (MHC) is one of the most gene-dense and polymorphic regions in the vertebrate genome. It plays an important role in the development and regulation of immune response in both adaptive and innate immunity. Despite the fact that the domestic pig (*Sus scrofa*) MHC has been well characterized, studies of closely related wild suid (Suidae) and distantly related peccary (Tayassuidae) species have never been attempted. However, it has been hypothesised that the emergence of the MHC non-classical class Ib genes occurred after the MHC classical class Ia genes appeared and after the divergence of suids from peccaries, implying that class Ib genes are not present in peccaries. To test this, domestic pig MHC primers were designed to amplify classical Ia and non-classical Ib genes. Amplicons were cloned and sequenced from ten suid species from Africa, Asia and Europe and three peccary species from the Americas. Preliminary analyses show MHC non-classical class Ib genes are also found in peccaries, contradicting the previous hypothesis for the emergence of these group genes. To further understand the variation within this immune region and reveal the ancestral sequence of the class Ia/Ib series, more MHC primers are being tested.

Poster 29. Phylogeography and population genetics of rock-wallabies in the Kimberley, WA

Sally Potter[1] David Taggart[1], Steve Cooper[1,2] and Mark Eldridge[3]

[1]School of Earth and Environmental Science and Australian Centre for Evolutionary Biology and Biodiversity, The University of Adelaide, South Australia, 5005 Australia. [2]Evolutionary Biology Unit, South Australian Museum, North Terrace, Adelaide, South Australia 5000. [3]Molecular Biology (Terrestrial Vertebrates), Australian Museum, College Street, Sydney, New South Wales 2010.

Three species of rock-wallabies are found in the Kimberley region of Western Australia, the short-eared rock-wallaby (*Petrogale brachyotis*), the Monjon (*P. burbidgei*) and the Nabarlek (*P. concinna*). These rock-wallabies form the *brachyotis* group within *Petrogale* and are the least understood (in terms of population genetics and ecology) of all 16 rock-wallaby species (Eldridge and Close 1997). Molecular genetic analyses will be used to investigate genetic diversity and gene flow amongst populations, as well as mating systems and social organisation. To date, 53 samples have been obtained from 15 populations including representatives of all three species. Initial mtDNA control region sequencing has revealed high levels of sequence divergence (~5%) amongst *P. brachyotis* populations sampled from the Kimberley. Microsatellite genotyping from ~15 markers will also be utilised to investigate the level of genetic diversity within populations, determine gene flow patterns, taxonomic classification and management units for conservation. This data will be used to assist in future management and conservation of rock wallaby populations.

Eldridge MBD, Close RL (1997) Chromosomes and evolution in rock-wallabies, *Petrogale* (Marsupialia: Macropodidae). *Australian Mammalogy* **19**, 123-135.

Poster 30. Mating and offspring frequencies under partial outcrossing in a structured population

Alan E. Stark

Balgowlah NSW, Australia 2093

This paper gives a model of a structured population with respect to an autosomal locus with two alleles. The population reproduces in discrete and non-overlapping generations. The population is assumed to be in equilibrium in that exactly the same distribution of genotypic proportions is reproduced in each generation. The population is subdivided into 'localities' which are characterized

by the local gene frequencies. Within each locality the genotypic proportions may depart from Hardy-Weinberg proportions and the same fixation index applies to all localities. The system departs from reality by assuming that the frequency of the first allele follows the beta distribution. However, this enables a convenient way to derive the mating frequencies of parents so that equilibrium is maintained. Wright's *F*-statistics are applied to characterize the population as a whole. The system is extended to permit an arbitrary level of outbreeding.

Stark AE (2008) Mating and offspring frequencies under partial outcrossing in a structured population. *Genet. Mol. Biol.*, 31:23-26.

Poster 31. Mitochondrial DNA analysis of Platypus from New South Wales

Amelia Swan[1], Chandramaya Siska Damayanti[1], Stephen Kolomyjec[2], Tom Grant[3], Chris Moran[1] and Jaime Gongora[1*]

*[1]Centre for Advanced Technologies in Animal Genetics and Reproduction, Faculty of Veterinary Science, RMC Gunn Building B19. University of Sydney, NSW 2006, Australia. [2]School of Marine and Tropical Biology, James Cook University, Townsville, QLD, Australia. [3]School of Biological, Earth and Environmental Sciences, University of New South Wales, Australia 2052. *Corresponding author: j.gongora@usyd.edu.au*

The platypus (*Ornithorhynchus anatinus*) is a monotreme endemic to Australia. Its current distribution is the streams and rivers of Queensland, New South Wales, Victoria 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 within the platypus is very limited. Here we analyse the genetic variation of the mitochondrial DNA control region (mtDNA CR) sequences of platypus from the Upper Shoalhaven, Upper Nepean, Cotter and Cotton Rivers in New South Wales: the mtDNA CR (~1kb) of the platypus was amplified by PCR and directly sequenced. Preliminary analysis shows that the mtDNA CR of representative individuals from these locations have low genetic variation (~0.02). More samples are currently being analysed to gain a further understanding of the phylogenetic relationships, contribution of maternal lineages and genetic diversity of the platypus in New South Wales. This information, in addition to other current studies from platypus from QLD, Victoria and Tasmania, will contribute to the assessment of the level of genetic differentiation within this species, assisting molecular ecology studies and conservation plans.

Poster 32. Cytogenetics of the *Simulium tuberosum* group (Diptera: Simuliidae) in Thailand.

Ubon Tangkawanit*, Chaliow Kuvangkadilok, Visut Baimai and Peter H Adler

Department of Biology, Faculty of Science, Mahidol University, Bangkok, 10400, Thailand.

The polytene chromosomes of 3,347 larvae of the *Simulium tuberosum* group collected from 59 locations in northern, northeastern, central and southern Thailand were analyzed. Band by band comparisons, relative to the established standard chromosome map for the subgenus Simulium, revealed 17 cytogenetically distinct taxa, based mainly on fixed, sex-linked and floating inversions in the long arm of chromosome III. Six of these taxa correspond to morphologically described species, i.e., S. doipuiense, S. rufibasis, S. setsukoae, S. tani, S. yuphae and S. weji; 2 unknown species (unknown sp.1 and unknown sp.2) were discovered. Two cytoforms (cytoform A and B) were found in S. doipuiense based on different sex-linked inversions, whereas 9 cytoforms (cytoforms A-I) were present in S. tani. S. tani cytoforms A and E differed from the standard cytoform (cytoform B) by fixed inversions. Cytoforms C, D, F, G and H were distinguished by sex-linked inversions, X_2Y_1, X_0Y_1, X_2Y_0, X_1Y_2 and X_3Y_0, respectively. Cytoform I was characterized by floating inversions. Shared unique chromosomal features, relative to the subgeneric standard chromosome map allowed evolutionary relationships among the cytotaxa to be inferred. Ecologically, the distributions of larvae of each taxon may be influenced by some macro- and microhabitat factors.

Poster 33. A HYBRID EVOLUTIONARY ALGORITHM FOR NUCLEOSOME POSITIONING

Armita Zarnegar[1], Mohammed M Islam[2], Peter Vamplew[3], Andrew Stranieri[1]

[1]School of Information Technology and computer science, University of Ballarat [2]School of Science, University of Ballarat

Eukaryotic DNA is packaged into arrays of Nucleosomes (chromosome) inside the cell nucleus. Nucleosomes appear to have two major

roles within the cell nucleus. First, they provide compaction to fit DNA strands into the cell nucleolus. Second, they play an important role in gene regulation by preventing the interaction of various DNA binding proteins with genomic DNA. Their second role makes Nucleosomes important for the purpose of controlling gene regulation as it affects the sequence accessibility. In over 20 years, there have been several studies for predicting the nucleosome positions. This is an interesting and challenging topic in Molecular biology today.

A recent research study demonstrated that the nucleosome positions have been encoded in the genomes using Dinucleotide sequences patterns can be used to predict ~50% of the Nucleosomes positions. In this study and previous works, mainly statistical methods have been used.

In this paper, our aim is to design a hybrid computational algorithm to recognize the motif in DNA sequences which has affinity with Nucleosome positions. We propose a hybrid Evolutionary algorithm called memetic algorithm, to combine the two aspects of the local search and global search. We found that this combined approach is compatible with the nature of the problem where we are looking to find the pattern of Dinucleotides inside each nucleosome region and also to find the nucleosome position along the genome. We applied this approach on a reliable dataset of Yeast DNA which has been previously used in a key research study in the field. Our result demonstrates a moderate prediction ability of DNA sequence for Nucleosome positioning. We also show that the usage of evolutionary techniques in this problem is as effective as statistical methods.

Poster 34. Molecular Genetics of Mitochondrial Function and Net Feed Efficiency in Cattle

Nadiatur Akmar Zulkifli, Madan Naik, Wayne Pitchford and Cynthia Bottema

School of Agriculture, Food and Wine, The University of Adelaide, South Australia, 5371 Australia

Net feed efficiency is an economically important trait in livestock. Net feed efficiency is affected by many factors including diet and genetics. Net feed efficiency of an animal depends on the ability of the animal to consume less feed than expected based on their weight gain and weight maintained during feed testing. This occurs by improving the utilisation of nutrients and energy from the feed for maintenance and growth. Recent work has implicated mitochondrial function as being important in net feed efficiency in livestock. The objectives of this study are to identify genes involved in mitochondrial function that may affect net feed efficiency in cattle based on QTL mapping and to study the relationship between these genes and mitochondrial function through biochemical assays. Several QTL affecting net feed efficiency were mapped in Jersey x Limousin backcross progeny. Based on the QTL, candidate genes involved in mitochondrial function have been selected. These candidate genes are being screened for DNA variants that might be used as DNA markers for selecting highly efficient animals.

Poster 35. Relationships between chromosome pairing and meiotic silencing of unpaired DNA (MSUD) in *Neurospora crassa*

William G. Alexander[1], Namboori B. Raju[2], Hua Xiao[1], Thomas M. Hammond[1], Tony D. Perdue[3], Edward G Barry[3], Robert L. Metzenberg[4], Jason E. Stajich[5], Patricia J. Pukkila[3], and Patrick K.T. Shiu[1]

[1]Division of Biological Sciences, U. Missouri, Columbia, MO, USA; [2]Dept. Biological Sciences, Stanford University, Stanford, CA, USA; [3]Dept. of Biology, U. North Carolina at Chapel Hill, Chapel Hill NC, USA; [4]Dept. of Biology, California State University, Northridge, CA, USA; [5]Dept. of Plant & Microbial Biology, U. California, Berkeley, CA, USA

N. crassa has an efficient mechanism to detect heterozygosities in the genome and to silence the expression of all copies of the heterozygous gene(s) during meiosis, a process known as MSUD. Previous work has shown that crosses lacking SAD-2 (an essential component of the silencing machinery) exhibit abnormal chromosome pairing, while crosses which self-silence *sad-2* exhibit normal meiotic progression. We have found that maternal and paternal sources of *sad-2* are silenced with equal efficiency. We have also examined the potential roles of additional genes in the silencing process. We conclude that *dcl-1* which encodes one of two known dicers in *N. crassa* is essential for silencing and also for meiosis, while *spo11* (which is known to be required for meiotic chromosome pairing in *N. crassa*) is not required for MSUD. We have also demonstrated colocalization of DCL-1 with SAD-1 (an RdRP), SAD-2, and SMS-2 (an Argonaute) in the perinuclear region.

Poster 36. Mitochondrial control region analyses of the Saltwater Crocodile from the Northern Territory.

Naomi Ratcliff[1], Chandramaya Siska Damayanti[1], Chris Moran[1], Sally Isberg[2], Jaime Gongora [1]*.

[1]Centre for Advanced Technologies in Animal Genetics and Reproduction, Faculty of Veterinary Science, RMC Gunn Building B19. University of Sydney, NSW 2006, Australia. [2]Porosus Pty Ltd, PO Box 86, Palmerston NT 0831, Australia

The saltwater crocodile (*Crocodylus porosus*) is one of the two species of crocodilians distributed across Australia. The saltwater crocodile is important in Australia ecologically and for the tourism and animal emerging industries. This study presents preliminary analyses of the mitochondrial DNA control region (mtDNA CR) sequence variation and phylogenetic relationships of more than 30 Australian saltwater crocodiles from six locations in the Top End of Australia's Northern Territory. CR sequences were aligned and compared with published saltwater crocodile sequences in GenBank from Queensland, Southeast Asia and the western Pacific Ocean, as well as with other crocodilian species. CR phylogenetic analyses show that saltwater crocodiles from Australia cluster in a separate clade from specimens from other countries. Pairwise genetic comparison of CR sequences show that the level of divergence within the saltwater crocodile is lower than that observed within the Central American Morelet's crocodile, similar to that observed in American Alligators and higher than that observed in captive Chinese alligators and the other crocodile species included. More samples are being analysed to provide further understanding of the genetic diversity within the Australian saltwater crocodile.

Poster 37. MiRNAs and piRNAs in platypus and echidna

Elizabeth P. Murchison[1*], Pouya Kheradpour[2], Ravi Sachidanandam[1], Carly Smith[1], Zhenyu Xuan[1], Frank Grützner[4], Alexander Stark[2,3], and Gregory J. Hannon[1]

*[1]Watson School of Biological Sciences, Howard Hughes Medical Institute, Cold Spring Harbor Laboratory, 1 Bungtown Road, Cold Spring Harbor, NY 11724, USA.[2]Computer Science and Artificial Intelligence Laboratory, Massachusetts Institute of Technology, Cambridge, MA 02139, USA. [3]Broad Institute of MIT and Harvard, Cambridge, MA 02141, USA . [4]Discipline of Genetics, School of Molecular & Biomedical Science, The University of Adelaide, 5005 SA, Australia. *Current address, Comparative Genomics Group, Research School of Biological Sciences, The Australian National University, Canberra 0200 ACT, Australia*

Small RNA pathways play evolutionarily conserved roles in gene regulation and in defense from pathogenic and parasitic nucleic acids. The character and expression patterns of small RNAs show conservation throughout animal lineages, but specific animal clades also show variations on these recurring themes, including species-specific small RNAs. The monotremes, with only platypus and four species of echidna as extant members, represent the basal branch of the mammalian lineage. Here, we examine the small RNA pathways of monotremes by deep sequencing of six platypus and echidna tissues. We find that highly conserved microRNA species display their signature tissue-specific expression patterns. In addition, we find a large rapidly-evolving cluster of miRNAs on platypus chromosome X1 which is unique to monotremes. Platypus and echidna testes contain a robust piRNA system which appears to be participating in ongoing transposon defense.

List of participants at 24 June 2008

First name	Surname	Organisation	Email
Kym	Abrams	University of Adelaide	kym.abrams@adelaide.edu.au
David	Adelson	University of Adelaide	david.adelson@adelaide.edu.au
Shahid	Ali	University of Melbourne	s.shahid@pgrad.unimelb.edu.au
Sara	Annan		sarannan@hotmail.com
Juie	Avis	Australian National University	u4302598@anu.edu.au
Sureshkumar	Balasubramanian	University of Queensland	mb.suresh@uq.edu.au
Eldon	Ball	Australian National University	eldon.ball@anu.edu.au
Lisa	Bardsley	University of Melbourne	l.bardsley2@pgrad.unimelb.edu.au
Stephen	Barker	University of Queensland	s.barker@uq.edu.au
Nigel	Beebe	University of Queensland	n.beebe@uts.edu.au
Katherine	Belov	University of Sydney	kbelov@vetsci.usyd.edu.au
Hannah	Bender	Australian National University	hannah.bender@anu.edu.au
Navdeep	Bhatti	University of Auckland	navdeep_66rn@yahoo.co.in
Michaela	Blyton	Macquarie University	michaela.blyton@students.mq.edu.au
Tessa	Bradford	University of Adelaide	tessa.bradford@adelaide.edu.au
Sarah	Bray	University of Adelaide	sarah.bray@adelaide.edu.au
William	Breed	University of Adelaide	bill.breed@adelaide.edu.au
Karen	Burke da Silva	Flinders University	karen.burkedasilva@flinders.edu.au
Bree	Buszard	Monash University	bree.buszard@sci.monash.edu.au
Corinne	Callegari	University of Adelaide	corinne.callegari@student.adelaide.edu.au
Sylvie	Callegari	University of SA	sylvie.callegari@postgrads.unisa.edu.au
David	Catcheside	School of Biological Sciences, Flinders University	david.catcheside@flinders.edu.au
Jackie	Chan	University of New South Wales	jackie.t.chan@gmail.com
Leiyao	Chang	University of Adelaide	lei.chang@student.adelaide.edu.au
Yuanyuan	Cheng	University of Sydney	yche8882@usyd.edu.au
Georgina	Cooke	Macquarie University	gcooke@bio.mq.edu.au
Alan	Cooper	University of Adelaide	alan.cooper@adelaide.edu.au
Ross	Crozier	James Cook University	ross.crozier@jcu.edu.au
Jack	da Silva	University of Adelaide	jack.dasilva@adelaide.edu.au
Tasman	Daish	University of Adelaide	tasman.daish@adelaide.edu.au
Brian	Dalrymple	CSIRO	brian.dalrymple@csiro.au
Janine	Deakin	Australian National University	janine.deakin@anu.edu.au
Paula	Dempsey		funnyhaha@e-access.com.au
Clio	Dersarkissian	University of Adelaide	clio.dersarkissian@adelaide.edu.au
Jenny	Donald	Macquarie University	jdonald@rna.bio.mq.edu.au
Stephen	Donnellan	South Australia Museum	donnellan.steve@saugov.sa.gov.au
Sally	Dunwoodie	Victor Chang Cardiac Research Institute	s.dunwoodie@victorchang.edu.au
Saman	Ebrahimi	University of Adelaide	saman.ebrahimi@adelaide.edu.au
Mark	Eldridge	Australian Museum	mark.eldridge@austmus.gov.au
Marianne	Frommer	University of Sydney	m.frommer@usyd.edu.au
Kioumars	Ghamkhar	CLIMA/UWA	kioumars@clima.uwa.edu.au
Gillian	Gibb	Massey University	g.c.gibb@massey.ac.nz
Nerida	Gibb	CSIRO Entomology	nerida.gibb@csiro.au
Philippa	Gibson	Australian National University	philippa.gibson@anu.edu.au
Frances	Goudie	Macquarie University	matcha3rose@gmail.com
Warwick	Grant	La Trobe University	w.grant@latrobe.edu.au
Jenny	Graves	Australian National University	jenny.graves@anu.edu.au
Stephen	Gregory	University of Adelaide	a1020177@adelaide.edu.au
Philippa	Griffin	University of Melbourne	p.griffin@pgrad.unimelb.edu.au
Frank	Grutzner	University of Adelaide	frank.grutzner@adelaide.edu.au
Jaro	Guzinski	Flinders University	guzi0002@flinders.edu.au
Jessica	Gwilliam	Macquarie University	jgwilliam@bio.mq.edu.au
Tom	Harrop	University of Melbourne	t.harrop@pgrad.unimelb.edu.au
Richard	Harvey	Victor Chang Cardiac	r.harvey@victorchang.edu.au

		Research Institute	
Susan	Harvey	Queensland University of Technology	sp.harvey@qut.edu.au
Simon	Ho	Australian National University	simon.ho@anu.edu.au
Gavin	Huttley	Australian National University	gavin.huttley@anu.edu.au
Reuben	Jacob	University of Adelaide	reuben.jacob@adelaide.edu.au
Weerachai	Jaratlerdsiri	University of Sydney	v_vet63@hotmail.com
Sanae	Jitkland	Mahidol University	g4436031student@mahidol.ac.th
Margaret	Katz	University of New England	mkatz@une.edu.au
Joan	Kelly	University of Adelaide	joan.kelly@adelaide.edu.au
Michael	Lardelli	SMBS, University of Adelaide	michael.lardelli@adelaide.edu.au
Kynan	Lawlor	University of Adelaide	kynan.lawlor@adelaide.edu.au
Martin	Lewis	University of Adelaide	martin.lewis@adelaide.edu.au
Yuan	Li	University of Adelaide	yuan.li@adelaide.edu.au
Brett	Lidbury	University of Canberra	brett.lidbury@canberra.edu.au
Shu Ly	Lim	University of Adelaide	shu.lim@adelaide.edu.au
Yinghong	Liu	University of Adelaide	yinghong.liu@adelaide.edu.au
Andrew	Lloyd	University of Adelaide	andrew.lloyd@adelaide.edu.au
Cintia	Martinez	La Trobe University	c6martinez@students.latrobe.edu.au
Robert	Mason	Australian Museum	rob.mason@austmus.gov.au
Oliver	Mayo	CSIRO	oliver.mayo@csiro.au
Elizabeth	McGraw	University of Queensland	e.mcgraw@uq.edu.au
Hilary	Miller	Victoria University of Wellington	hilary.miller@vuw.ac.nz
Judith	Mitchell	University of Melbourne	j.mitchell2@pgrad.unimelb.edu.au
Amir	Mohammadi	Australian National University	amir.mohammadi@anu.edu.au
Elizabeth	Murchison	Australian National University	murchiso@cshl.edu
Veronica	Murtagh	Australian National University	veronica.murtagh@anu.edu.au
Steven	Myers	Flinders University	myer0026@flinders.edu.au
Yeap Shen	Ng	University of Adelaide	yeap.ng@adelaide.edu.au
Fred	Nijhout	Duke University	hfn@duke.edu
Svanhild	Nornes	University of Adelaide	svanhild.nornes@adelaide.edu.au
Irida	Novianti	University of Adelaide	irida.novianti@student.adelaide.edu.au
Louise	O Keefe	University of Adelaide	louise.o'keefe@adelaide.edu.au
Denis	O'Meally	Australian National University	denis.omeally@anu.edu.au
Shoko	Okada	CSIRO Division of Entomology	shoko.okada@csiro.au
Kym	Ottewell	University of Adelaide	kym.ottewell@adelaide.edu.au
Pornpimol	P.Rattanatrai	University of Sydney	ppue2466@usyd.edu.au
Hardip	Patel	Australian National University	hardip.patel@anu.edu.au
Vidushi	Patel	Australian National University	vidushi.patel@anu.edu.au
Charles	Pearce	University of Adelaide	charles.pearce@adelaide.edu.au
David	Penny	Massey University	d.penny@massey.ac.nz
Matt	Phillips	Australian National University	matt.phillips@anu.edu.au
Amanda	Potter		pottsy1000@hotmail.com
Sally	Potter	University of Adelaide	sally.potter@adelaide.edu.au
Luke	Price	University of Adelaide	luke.price@adelaide.edu.au
Patricia	Pukkila	University of North Carolina	Pukkila@bio.unc.edu
Nic	Rawlence	Australian Centre for Ancient DNA	nicolas.j.rawlence@adelaide.edu.au
Emily	Remnant	University of Melbourne	e.remnant@pgrad.unimelb.edu.au
Tamarind	Reynolds	Macquarie University	tamarind_reynolds@hotmail.com
Allen	Rodrigo	University of Auckland	a.rodrigo@auckland.ac.nz
Elizabeth	Ross	Latrobe University	emross@students.latrobe.edu.au
Robert	Saint	Australian National University	robert.saint@anu.edu.au
Beth	Schlotfeldt	Flinders University	schl0030@flinders.edu.au
Jeremy	Shearman	University of New South Wales	j.shearman@student.unsw.edu.au
Anna	Sheppard	University of Adelaide	anna.sheppard@adelaide.edu.au
William	Sherwin	University of New South Wales	w.sherwin@unsw.edu.au
Mark	Sistrom	South Australian Museum	mark.sistrom@adelaide.edu.au
David	Smyth	Monash University	david.smyth@sci.monash.edu.au

John	Sved	University of Sydney	jsved@usyd.edu.au
Amelia	Swan	University of Sydney	aswa2617@usyd.edu.au
Tamar	Sztal	University of Melbourne	t.sztal@pgrad.unimelb.edu.au
Ubon	Tangkawanit	Mahidol University	g4436030@student.mahidol.ac.th
Vicki	Thomson	University of Adelaide	vicki.thomson@adelaide.edu.au
Rugang	Tian	University of Adelaide	rugang.tian@adelaide.edu.au
Jeremy	Timmis	University of Adelaide	jeremy.timmis@adelaide.edu.au
Richard	Todd	Melbourne University	rbtodd@unimelb.edu.au
Paulina	Truong	Monash University	ptru3@student.monash.edu.au
Ben	Tucker	University of Adelaide	ben.tucker@adelaide.edu.au
Clare	van Eyk	University of Adelaide	clare.vaneyk@adelaide.edu.au
Tatiana	Vassilieva	Australian National University	tatiana.vassilieva@anu.edu.au
Claire	Walker	University of Adelaide	claire.walker@student.adelaide.edu.au
Coral	Warr	Monash University	coral.warr@sci.monash.edu.au
Camilla	Whittington	University of Sydney	c.whittington@usyd.edu.au
Ken	Wolfe	Trinity College	khwolfe@tcd.ie
Henry	Ye	University of Queensland	s4062139@student.uq.edu.au
Nadiatur Akmar	Zulkifli	University of Adelaide	nadiatur.zulkifli@student.adelaide.edu.au