

Tuesday 29th July

9.30 – 12.30 Diagnostic Laboratories and the HVP Consortium

13.00 – 15.00 Registration, poster mounting and trade set-up with coffee

15.00 – 15.05 Welcome Address
Richard Cotton

Session 1 **Clinical Molecular Genetics**
Chair: Richard Cotton

15.05 - 15.35 Zbigniew Rudzki
The role of mutation detection in determining the most appropriate kinase inhibitor therapy for patients with chronic myeloid leukaemia.

15.35 - 16.05 Dean Phelan
The laminopathies mystery: mutation detection is not enough!

16.05 – 16.35 Caitlin Edwards
Effectiveness of an extended mutation detection strategy in NBS for CF in Western Australia

16.35 - 17.05 COMPANY LECTURE

SEQUENOM
Darryl Irwin
Medical Applications of Nucleic Acid Mass Spectrometry

17.05 – 17.30 *Coffee break*

17.30 - 18.30 PLENARY LECTURE

Steve Sommer
The Epidemiology Vision of Clinical Molecular Diagnosis in the Era of Massively Parallel Sequencing

18.30 - 19.00 *Welcome drinks at sunset on Charter's Lawn*

19.00 *Welcome dinner - BBQ on Charter's Lawn*

Wednesday 30th July

8.00 - 8.30 *Registration with coffee on arrival*

8.30 - 9.30 PLENARY LECTURE

Susan Forrest
Progress and Plans at the AGRF

Session 2 **Mutation Detection – Disease I**
Chair: Serge Kovalenko

9.30 – 9.50 Joyce Teo
Mutation scanning in the breast cancer susceptibility gene PALB2: are some female carriers at high risk of breast cancer?

9.50 – 10.10 Chelsee Hewitt
High resolution melting KRAS exon 2 mutation detection; from research to diagnostics

10.10 – 10.40 COMPANY LECTURE

ILLUMINA
Dan Headon
Technology review: application of the latest genomic tools to cancer research

10.40 - 11.10 *Coffee break*

Session 3 **Mutation Detection – Disease II**
Chair: Zbigniew (Barney) Rudzki

11.10 – 11.40 Lai Poh San
High-resolution melt (HRM) analysis for genotyping mutations in Duchenne Muscular Dystrophy

11.40 – 12.00 Deborah Norman
Development of MUTYH genetic test in colorectal cancer

12.00 – 12.20 R. Dubey
Variant detection in Charge syndrome

12.20 – 12.50 COMPANY LECTURE

SHIMADZU
John Hewetson
A fully automated microchip electrophoresis system for DNA and RNA size analysis

12.50 – 14.00

Lunch

Session 4

Methods I

Chair: Val Hyland

14.00 - 14.20

Elizabeth Algar

Application of methylation sensitive high resolution melting in the diagnosis of growth disorders

14.20 – 14.40

Shannon Cowie

The long and the short of sequencing: time, cost and reliability!

14.40 – 15.00

Thomas Dantoft

Use of high-resolution melting analysis for the detection of unknown changes in DNA

15.00 – 15.30

COMPANY LECTURE

APPLIED BIOSYSTEMS

Mike Payne

The Solid Sequencer – Towards Detection Of Whole Genome Variation

15.30 – 16.00

Coffee break

Session 5

Methods II

Chair: Cyril Mamotte

16.00 – 16.30

Desirée du Sart

Validation of the new kid in diagnostics: genotyping with maldi-tof mass spectrometry

16.30– 17.00

Edward Edkins

Use of stimulated white blood cells in mutation detection

17.00 – 17.20

Elizabeth Tindall

Assessing high resolution melt curve analysis for accurate detection of gene variants in complex DNA fragments

17.20 – 18.30

Poster Session & Trade

19.00

*Gala dinner – Matso's Broome Brewery
Meet in foyer 18.45*

Thursday 31st July

Session 6

MOLECULAR GENETICS SOCIETY OF AUSTRALASIA
Chair: Desirée du Sart

8.30 - 9.00

PLENARY LECTURE

Steve Sommer

PAP/MAP for Monitoring of Therapy or Recurrence in Breast Cancer

9.00 - 9.30

Michael Abdo

Clinical molecular genetics in Western Australia

9.30 - 10.00

John Macmillan

Addressing the weak link in annotating the human genome and its phenotypes, the busy clinician

10.00 - 10.30

Val Hyland

Triplet repeat pathogenic mutations in the DMPK, DRPLA, FMRI, FXN, HTT, PABPN1, SCA1, SCA2, SCA3, SCA6 and SCA7 genes detected in a diagnostic molecular genetics laboratory

10.30 - 11.00

Coffee break

11.00 - 11.30

Jonas Bjorkman

Unclassified variants in familiar colorectal cancer in Queensland account for 30 percent of the total number of mutations identified

11.30 - 12.30

Discussion workshop
Pathogenic or not?

12.30 - 13.15

Lunch

13.15 - 17.00

Excursion to Willie Creek pearl farm

17.00 - 18.00

Sunset at Cable Beach & Free time

18.00 - 21.00

BBQ on Cable Beach

Friday 1st August

Session 7

Methods III

Chair: Vanessa Hayes

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| 8.30 – 8.50 | Melanie Smith
MLPA or real time for SMA population screening |
| 8.50 – 9.10 | Julie Stampalia
cDNA sequence analysis and IHC identify pathogenic sequence variants in the PMS2 gene |
| 9.10 – 9.30 | Elena Takano
Optimisation and quality assessment of DNA extraction from formalin fixed paraffin embedded tissue for mutation detection |
| 9.30 – 9.50 | Justin Hasell
What, no more radio-isotopes?? |
| 9.50 – 10.10 | Anna Ritchie
The critical step of MLPA |
| 10.10 – 10.30 | Trent Burgess
Detection of deletions of the ITPR1 gene in patients with autosomal dominant cerebellar ataxias using a home grown MLPA assay |
| 10.30 – 10.50 | Vanessa Calabro
What is the value of RNA analysis; can we report an RNA only change? |
| 10.50 – 11.20 | <i>Coffee Break</i> |

Session 8

Mutation Detection IEM & Database I

Chair: David Ravine

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| 11.20 - 11.50 | Richard G.H. Cotton
Pilot studies of the Human Variome Project |
| 11.50- 12.10 | Bruce Bennetts
Female heterozygous ornithine transcarbamylase (OTC) deficiency patient - large deletion identified by MLPA and mapped by CGH microarray |
| 12.10 – 12.30 | Sarah Pantaleo
Testing for mitochondrial DNA deletions in the laboratory |
| 12.30 – 12.50 | Lin Rigby
A thermal melt analysis assay to detect COL4A3 and COL4A4 mutations in thin basement membrane nephropathy |

12.50 – 13.10	Jamie - Lee Ricciardi Mutation detection in skeletal dysplasia
13.10 – 13.30	Shawn Jones Can LOVD be used to collect laboratory and clinical data in a diagnostic/clinical environment
13.30 – 13.40	<i>Wind up / next meeting - R.G.H. Cotton</i>
13.40	<i>Meeting Ends – Lunch</i>

Poster Presentations

Poster 1	Genetic alterations of <i>p53</i> in brain astrocytoma A. Al-Shabeeb
Poster 2	Cystic fibrosis mutation screening using the XTAG TM cystic fibrosis kit A. Duffy
Poster 3	Haemophilia a diagnostic testing in Victoria M. Gavrilu
Poster 4	Are superseded techniques really inferior in the molecular diagnosis of juvenile HD? M. Kaps
Poster 5	Automation of BRCA express sequencing method S. Macaskill
Poster 6	The detection of MELAS m.3243A>G: urine epithelial cells versus skeletal muscle, blood and hair follicles R. Marotta