

Molecular Genetics Society of Australasia Meeting

18.00 Sunday 1st August – 15.30 Tuesday 3rd August

Tuesday 3rd August

13.30 – 15.45 Registration & Poster Mounting

16.00 – 16.05 Welcome Address

Richard Cotton

Session 1 : The Australian Node of The Human Variome Project

Chair: John Macmillan

16.05 – 16.25 Progress and Pilots of the Human Variome Project

Richard Cotton

16.25 – 16.45 Data, Systems and Ethics – Designing a Country Node for the Human Variome Project

Alan Lo

16.45 – 17.05 Building MAWSON – challenges in clinical genetic databases

Andrew Dubowsky

17.05 – 17.25 The Human Variome Project and the Diagnostic Laboratory: The Ideal Need

Desirée Du Sart

17.25 – 17.45 Defining Curation

Timothy Smith

17.45 – 18.05 Gathering and curating retrospective genetic data from diagnostic labs: the French UMD-*DMD* and UMD-*CFTRbases*

Mireille Claustres

18.05 – 18.30 Discussion

19.00 -

Welcome Reception & Roaming Dinner

Quolls, Greygums & Brushtails areas of hotel

Wednesday 4th August

Session 2: Plans and Analyses

Chair: Desireé Du Sart

9.00 – 10.00

KEYNOTE SPEAKER

French national plan for rare diseases.
The lab's perspective

Mireille Clausters

10.00 – 10.20

Mutational analysis of TSC1 and TSC2 genes in a Western Australian cohort

Deborah Norman

10.20 – 10.40

Honing In On Hydrocephalus: Screening The LICAM Gene

Ratna Dubey

10.40 – 11.10

Coffee Break & Trade

SESSION 3: Using size and mass to detect variants

Chair: Val Hyland

11.10 – 11.30

Use of nanoparticles in detection of single nucleotide mutations for clinical diagnostics

Lai Poh San

11.30 – 11.50

MALDI-TOF Mass spectrometry in clinical diagnosis – The Cystic Fibrosis story

Sarah Pantaleo

11.50 – 12.10

MALDI-TOF Mass spectrometry in diagnostic screening for Ashkenazi Jewish diseases

Melissa Chow

12.10 – 12.40 **COMPANY LECTURE**

SEQUENOM

High throughput oncogene mutation profiling using MALDI-TOF Mass Spectrometry

Darryl Irwin

12.40 – 14.00 Lunch – Quolls Restaurant

SESSION 4: Quality Control & Screening for Mutations

Chair: Bruce Bennetts

14.00 – 14.50 An international approach to quality assurance in molecular genetics –
The European Molecular Genetics Quality Network and EUROAGENTEST

Rob Elles

14.50 – 15.10 Identification of somatic mutations in patients with metastatic melanoma

Karen Ambler

15.10 – 15.30 The contribution of congenital CMV infection and mutations in the GJB2,
SLC26A4, and mtDNA 12S rRNA genes to hearing loss in Australian children
(the LOCHI study)

Henrik Dahl

15.30 – 16.00 **COMPANY LECTURE**

ILLUMINA

Challenging Diseases – How Illumina is revolutionising mutation detection
with next generation array and sequencing technologies

Arjuna Kumarasuriyar

16.00 – 17.00 Coffee Break & Poster Session/Trade

17.30 – 19.00 Wildlife Spotighting Tour

Meet promptly in Foyer, dress warm with coat, hat etc.

19.00 - Dinner in the Grey Gums Restaurant of the hotel

Thursday 5^h August

Session 5: How much is enough? Persistence pays

Chair: Jenny Leary

- 8.30 – 8.50 The Long Way home: An FAP Cold Case solved!
Shannon Cowie
- 8.50 – 9.10 Insert/delete Friedreich ataxia here?
Justin Hassell
- 9.10 – 9.30 White blood cells detect MLH1 promoter methylation and no germline
mutation in two siblings with HNPCC
Julie Stampalia
- 9.30 – 10.00 **COMPANY LECTURE**
QIAGEN
Pyrosequencing: Sequence Based Detection and Quantification
Andrea A Tesoriero

10.00 – 10.30 Coffee Break & Trade

SESSION 6: Creating Variants in Mice & Wheat

Chair: Andrew Fellowes

- 10.30 – 10.50 A recombination hotspot leads to sequence variability within a novel gene and
contributes to complex disease susceptibility
Tom Brodnicki
- 10.50 – 11.10 Sleeping Beauty transposon mutagenesis in NOD mice
Colleen Elso
- 11.10 – 11.30 SNP detection in hexaploid wheat
Chongmei Dong

SESSION 8: Splicing and Discussion

Chair: Richard Cotton

11.00 – 11.20 Empirical validation of DNA sequence variants predicted to encode aberrant RNA splicing

Andrew Dubowsky

11.20 – 11.45 General Discussion

MEETING ENDS

11.45 – 12.45

Lunch – Quolls Restaurant

12.45 Bus departs for Launceston airport PROMPTLY

10.00 Saturday 7th August - Bus departs for Launceston airport PROMPTLY