

Sunday 1st August

18.00 – 19.30 **Welcome Reception & Registration**
Brushtails Bar (make own arrangements for dinner)

Monday 2nd August

Session 1 **Laboratory Updates**
Chairs: Kathie Friend & Peter Taylor

8.30 – 10.00 **5 minute presentations from each laboratory**

10.00 – 10.15 **SPONSORED COMPANY LECTURE**

ABBOTT MOLECULAR

Development of A High-Throughput Assay For Detecting Fragile X Expanded Alleles

Tom Berkovits

10.15 – 10.45 **Coffee Break**

Session 2 **Keynote Speaker**
Chairs: Val Hyland & Tiffany Wotton

10.45 – 11.30 **MGSA INVITED SPEAKER**

Fragile X Syndrome Newborn Detection: Results from a South Carolina Pilot Study

Charles Schwartz

Session 3 **Clinical Update**
Chairs: Mike Field & Carolyn Rogers

11.30 – 11.40 **Are neurocognitive impairments associated with small or intermediate expansions on the *FMRI* gene?**

Kim Cornish

11.40 – 11.50 **Small CGG Repeat Expansions in the Fragile X Gene Contribute to Parkinsonism**

David Godler

- 11.50 – 12.00 **Proposed Clinical Guidelines For Fragile X Testing**
Carolyn Rogers
- 12.00 – 12.10 **Referrals patterns and testing for the *FMR1* gene in Victoria for the last ten years**
Sylvia Metcalfe
- MGSA POSTERS INTRODUCTION**
- 12.10 – 12.12 **Improved Methodology for Assessment of mRNA Levels in Blood of Patients with FMRI Related Disorders**
David Godler
- 12.12 – 12.14 **An Improved Method For The Detection Of CGG Trinucleotide Repeats In Fragile X Syndrome**
Rick Woods
- 12.14 – 12.16 **One cell versus two cell biopsy for embryos undergoing Preimplantation Genetic Diagnosis (PGD) with translocation testing**
Danni Brodie
- 12.16 – 12.18 **An allele specific, real-time method for detecting MTHFR mutation c.1298A>C using High Resolution Melt Curve analysis**
Josephine Carpenter
- 12.18 – 12.20 **Microsatellite madness - problems during optimisation of multiplex QF-PCR for prenatal analysis of aneuploidy**
Kim Flintoff
- 12.20 – 12.22 **Legal aspects of medical malpractice in prenatal diagnosis**
Jacob Barg
- 12.22 – 12.45 **Discussion**
- 12.45 – 14.45 **Lunch – Quolls Restaurant**
&
Free Time
- Session 4** **General Molecular Genetics**
Chairs: Ted Edkins & Jenny Leary
- 14.45 – 14.55 **Somatic Mosaicism of a Novel Mutation within the *NEMO* Gene in a Family with Incontinentia Pigmenti**
Evelyn Douglas
- 14.55 – 15.05 **Quantification of mitochondrial mutation mutant load using High Resolution Melt analysis**
Desiree Du Sart
- 15.05 – 15.15 **Factors which impact on the quantification of copy number**
Justine Marum

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15.15 – 15.25 How Essential Is Cascade Testing For Determining Mutation Pathogenicity
Dean Phelan

15.25 – 15.35 A PALB2 Mutation Associated With High Risk Of Breast Cancer
Melissa Southey

15.35 – 15.45 A Case of Somatic Mosaicism for Long QT Syndrome with Transmission to Two Clinically Affected Offspring
Louisa Sanchez

15.45 – 16.00 SPONSORED COMPANY LECTURE
ILLUMINA

Redefining the Trajectory of Genetic Pathology. Next Generation Analysis Tools from Illumina: *Limitless applications harnessed with intensity and implemented with imagination.*

Tyrrell, Vanessa

16.00 – 16.30 Coffee Break & Poster Session

Session 5 Screening
Chairs: Bruce Bennetts & Sylvia Metcalfe

16.30 - 16.40 Community views of population carrier screening for fragile X syndrome
Alison Archibald

16.40 – 16.50 Acceptability and attitudes towards newborn screening for fragile X syndrome in postnatal mothers
Louise Christie

16.50 – 17.00 Newborn screening for the fragile X syndrome. Now or never?
Mike Field

17.00 – 17.10 Offering carrier screening for fragile X syndrome to non-pregnant women
Melissa Martyn

17.10 – 17.20 The Reliability and Feasibility of Newborn Screening for Fragile X Syndrome using a DNA Based First Tier Assay

Tiffany Wotton

17.20 – 18.30 Discussion

19.00 - Departure for Dinner at
Highland Restaurant - Cradle Mountain Lodge
Assemble in foyer for coach transfer & prompt departure

Tuesday 3rd August

Session 6 **Keynote Speaker**
Chairs: Val Hyland & Melissa Southey

8.30 – 9.15 **MGSA INVITED SPEAKER**
Next Generation Genetic Services in the UK and Europe
Rob Elles

Keynote Speaker
Chairs: Val Hyland & Melissa Southey

9.15 – 9.30 **MUTATION DETECTION INVITED SPEAKER**
Our experience with PGD for single gene disorders: cystic fibrosis as an example
Mireille Claustres

Session 7 **Fragile X Method development**
Chairs: Desirée du Sart & Andrew Fellowes

9.30 – 9.40 **Methylation of Novel Markers of Fragile X Alleles is Inversely Correlated with FMRP and FMRI Activation Ratio**
David Godler

9.40 – 9.50 **Detection of mosaicism at the FMRI locus**
Kathie Friend

9.50 – 10.00 **Novel epigenetic predictors of cognitive impairment in carriers of expanded FMRI alleles raise the possibility of a PCR-only workflow for FMRI diagnostic testing and population screening**
David Godler

10.00 – 10.30 **Discussion**

10.30 – 11.00 *Coffee Break*

Session 8 **Best Practice Guideline I**
Chairs: Bruce Bennetts, Kathie Friend, Val Hyland, David Ravine

11.00 – 12.30 **Fragile X Best Practice Document**
Discussion I

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12.30 – 14.30 *Lunch – Quolls Restaurant
& Free Time
(those departing on bus to airport leave here)*

Session 8 **Best Practice Guideline 2**
Chairs: Bruce Bennetts, Kathie Friend, Val Hyland, David Ravine

14.30 – 15.30 **Fragile X Best Practice Document**
Discussion 2

15.30 **MGSA Meeting ends**

15.30 – 16.00 *Coffee Break*

16.00 **8th Australasian Mutation Detection Meeting begins**