11th Australasian Mutation Detection Meeting: methods, genome sequencing - detecting variants one-by-one or genome-wide, cancer gene analysis & diagnostic applications

11th – 14th September 2016
Alice Springs, NT

PROGRAM

Subject to rearranging

Sunday 11th September	
12.00 – 13.00	**Registration** - foyer outside meeting room
13.00 – 14.00	**Lunch & Registration**
14.00 – 14.05	**Welcome Address** Desiree Du Sart *VCGS, Melbourne, VIC*
Session 1	**ANCIENT DNA**
14.05 – 15.05	**KEYNOTE SPEAKER** **Ancient DNA (exact title TBA)** Speaker from Prof. David Lambert's Lab, TBA *Ancient DNA Lab, Griffith University. QLD*
15.05 – 15.15	**Open Discussion**
Session 2	**FORENSIC DNA**
15.15 - 16.15	**KEYNOTE SPEAKER** **DNA in Forensics: its application to human identification and more** Dadna Hartman *Molecular Biology, Victorian Institute of Forensic Medicine, VIC*
16.15 - 16.25	**Open Discussion**
16.25 - 16.45	**Coffee Break**
Session 3	**NEXT STEPS FOR NATIONAL DATA SHARING - HVPA**
16.45 - 18.30	**Human Variome Project Australian Node. Details to come soon.** The Human Variome Project Australian Node is a national data sharing facility for improving clinical genetic testing services and supporting medical research.http://www.hvpaustralia.org.au/
18.30 - 19.00	**Free Time**

19.00 – 21.30	**Welcome Dinner – *Poolside - DoubleTree by Hilton***

Monday 12th September

Session 4	**LIQUID BIOPSY**
8.30 – 9.30	**KEYNOTE SPEAKER** **Circulating tumour DNA as a non-invasive diagnostic tool in cancer** Stephen Wong *Molecular Biomarkers and Translational Genomics Lab, Peter MacCallum Cancer Centre, VIC*
9.30 – 9.50	**Open Discussion**
9.50 – 10.10	**COMPANY LECTURE** **AGENA BIOSCIENCE** **How Agena Bioscience is Driving Down Limit of Detection for Solid Tumours and Liquid Biopsies** Yvette Emmanuel *Applications and Technology Scientist at Agena Bioscience Inc.*
10.10 – 10.20	**Rapid Fire Poster Presentations 1**
10.20 – 10.50	**Coffee Break**
Session 5	**NON-INVASIVE PRENATAL TESTING**
10.50 – 11.50	**KEYNOTE SPEAKER** **Whole genome NIPT: A window into the earliest stages of human development** Mark Pertile *VCGS, Melbourne, VIC*
11.50 – 12.20	**IVF and NIPT - the perfect match?** Peter Field
12.20 – 12.50	**Open Discussion**
12.50 – 13.50	**Lunch**
Session 6	**EXOME SEQUENCING**
13.50 – 14.50	**KEYNOTE SPEAKER** **Diagnostic Genome Analysis of Rare Hereditary Disorders** Hans Schaeffer *Radbourg Univ. Medical Center, Nijmegen, The Netherlands*

14.50 - 15.00	**Open Discussion**
15.00 – 15.20	**Analytical Validation of Exome Sequencing** Claire Love
15.20 - 15.40	**Clinical Exome Sequencing at VCGS** Dean Phelan
15.40 – 15.50	**Rapid Fire Poster Presentations 2**
15.50 – 16.20	**Poster Session (*poster presenters to stand by their poster*)**
Session 7	**EXTERNAL QUALITY ASSESSMENT FOR NGS TECHNOLOGIES**
16.20 - 17.30	**KEYNOTE SPEAKER** **EQA for NGS technologies** Desiree du Sart for European Molecular Genetics Quality Network (EMQN), UK
17.30 - 18.30	**Open Discussion**
18.30 -	**Evening at leisure**

Tuesday 13th September

Session 8	**WHOLE GENOME SEQUENCING DIAGNOSTIC NGS & NOVEL METHODS**
8.30 - 9.30	**KEYNOTE SPEAKER** **Whole Genome Sequencing Service at the Kinghorn Centre (exact title TBA)** Marcel Dinger
9.30 - 9.40	**Open Discussion**
9.40 - 10.00	**Hi-Plex for simple, accurate and low-cost mutation detection** Daniel Park
10.00 - 10.20	**Quantitative JAK2 V617F Assay Using Droplet Digital PCR** Geraldine Duncan
10.20 - 10.40	**COMPANY LECTURE** **BIO-RAD** **Drop Everything - Precision Biology with Droplet Digital™ PCR** Richard Harrison *Genomics Marketing Manager, Asia Pacific, Bio-Rad Laboratories Pty Ltd*
10.40 - 11.10	**Coffee Break**

Workshop 1	**VARIANT CURATION**
11.10 – 13.00	Panel • Marcel Dinger • Desiree du Sart • Scott Grist • Vince Harley (to be confirmed) • Hans Schaeffer Variant Curation • The general process followed by most labs • Cases • Live Curation The audience is asked to participate with open discussion
13.00– 14.00	**Lunch**
Session 9	**MUTATION DETECTION METHODS & APPLICATIONS**
14.00 - 14.20	**COMPANY LECTURE** **QIAGEN** **GeneReader - (title TBA)** Speaker TBA
14.20 - 14.40	**Y Chromosome microdeletions and Next Generation Sequencing** Peter Field
14.40 – 15.00	**Pitfalls of CALR mutation testing: Is there still a role for bone marrow biopsy in the diagnosis of myeloproliferative neoplasms?** Mary Koleth
15.00 - 15.20	**Sudden death and family cascade testing- expect the unexpected** Daniel Flanagan
15.20 - 15.30	**Gene mutations in myelodysplastic syndrome (MDS): a preliminary study from a Sydney South West Laboratory** Anne-Marie Marivel
15.30 - 15.40	**Using a Sequenom assay to detect mitochondrial mutant load in prenatal testing for mitochondrial disorder, NARP** Chelsea Holt
15.40 - 15.50	**Sensitive and specific colorimetric DNA detection by loop-mediated isothermal amplification coupled with invasive reaction assisted nanoparticles** Xueping Ma

15.50 - 16.00	**INNOVATION ZONE** 2 minute elevator pitch of a great idea, trend, tool, technique you developed/heard of etc...You do not have to be a company - Don't be shy! ○ TBA – is this you?
16.00 – 16.30	**Coffee Break**
16.30 – 17.00	**Free Time to get your outback gear on!**
17.00 – 22.30	**Dinner in the Outback** ***Assemble in the hotel foyer at 17.00 pm for prompt departure*** ***Dress in Outback style, jeans etc. Bring a jacket. Wear comfortable shoes!*** ***Bring sunglasses.***

Wednesday 14th September

Workshop 2	**POP UP DISCUSSION**
8.30 – 10.30	**YOU determine the topics** Please come to the meeting with one or more topics or issues that you feel should be discussed. The theme(s) for this workshop shall be determined by popular demand at the meeting. Delegates will be emailed before the meeting with a list of topics to think about.
10.30 – 10.40	**Windup & Summary** **MEETING END**
10.40 – 11.10	**Coffee Time – (*Lunch bags will be provided for you to take away)***