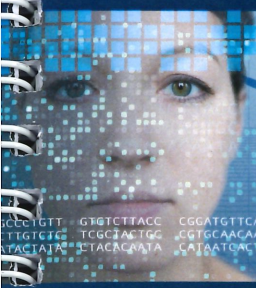

THE CHANGING FACE OF GENETIC MEDICINE

HGSA 38th Annual Scientific Meeting
Hilton Adelaide, South Australia
3-6 August 2014

MEETING HANDBOOK

HUMAN GENETICS SOCIETY OF AUSTRALASIA

Invited Speakers - International Speakers

Anna Middleton, Wellcome Trust Sanger Institute, Cambridge, UK

Dr Anna Middleton has had two parallel careers – the first as a practicing genetic counsellor, the second as a senior social scientist exploring the impact of genetic technology on people. She currently works at the Wellcome Trust Sanger Institute in Cambridge, UK; as the only social scientist on campus she leads an an international, mixed methods research project which explores attitudes towards sharing incidental findings from sequencing studies. Anna's PhD in Genetics and Psychology (awarded in 2000) was the first research to show that Deaf adults may use pre-natal genetic testing for deafness with a preference for having deaf children. She is a previous vice-chair of the Genetic Counsellor Registration Board (UK and ROI) and a current committee member of the Association of Genetic Nurses and Counsellors. She played a leading role in the creation of British genetic counselling policy on (i) the registration of overseas genetic counsellors in the UK (ii) the accreditation of MSc genetic counselling courses, (iii) the place of counselling supervision in registration and practice. Anna has over 80 published peer review papers, book chapters and spoken conference presentations. She has also edited two books on genetic counselling practice. Anna has a visiting fellowship at the Public Health Genomics Foundation in Cambridge, UK and as part of this contributes to the 'Realising Genomics' think tank which influences policy about the use of genomic technology in clinical practice. She also holds an honorary research position at the Ethox Department, University of Oxford; in 2011 she was awarded a visiting fellowship at the Brocher Foundation in Geneva, Switzerland.

Maximilian Muenke, NIH, USA

Dr. Max Muenke serves as the Chief of the Medical Genetics Branch of the Division of Intramural Research at the National Human Genome Research Institute, National Institutes of Health (NIH) in Bethesda, Maryland, USA and the Director of the NIH Medical Genetics and Genomic Medicine Residency and Fellowship programs. Dr. Max Muenke trained in Pediatrics in his native Germany. He pursued postdoctoral training in Human Genetics at Yale and the University of Pennsylvania and completed a fellowship in Clinical Genetics at the Children's Hospital of Philadelphia. For the past two decades, the focus of his research has been on the delineation and identification of the underlying causes of craniofacial anomalies in humans. His lab made seminal discoveries in linking Sonic Hedgehog signaling to normal and abnormal brain development in humans and has identified over a dozen genes involved in holoprosencephaly, the most common anomaly of the developing forebrain. His group identified several genes important in craniofacial disorders including the most common craniosynostosis syndrome, now termed Muenke syndrome. More recently, his lab has identified susceptibility loci for the most common behavioral disorder in childhood, Attention-Deficit/Hyperactivity Disorder (ADHD), with further research focused on predicting severity, treatment response, and long-term outcome. Dr. Muenke is also interested in personalized medicine, from understanding rare and common diseases to their treatment and prevention.

Detailed Program

Monday 4th August 2014

0730-1830	**REGISTRATION DESK OPEN**	*Foyer, Level 1*
0830-1100	**Plenary and Themed Session A** *Co-Chairs: Jan Hodgson and Clara Gaff* **Plenary 1: What's the fuss about incidental findings? Opportunistic screening and international attitudes** *Anna Middleton* **Plenary 2: Communicating about genetics: A family affair** *Carma Bylund* **Plenary 3: Development and implementation of clinical genomics** *John Mattick*	*Ballrooms A&B*
1100-1130	**MORNING TEA AND EXHIBITION**	*Foyer, Level 1*
1130-1300	**Plenary and Themed Session B** *Chair: Sue Thompson* **Plenary 4: Fatty acid oxidation defects: Phenotypes, outcome and the diagnostic dilemma** *Ute Spiekerkoetter* **Plenary 5: Newborn Screening and the Interface with treatment** *Bridget Wilcken*	*Ballrooms A&B*
1300-1400	**LUNCH AND EXHIBITION and Poster Viewing**	*Level 1/Balcony 1&2*
1400-1500	**CONCURRENT SESSIONS**	

1400-1500	**CONCURRENT 1: AACG** *Chair: Michael Gabbett* *Balcony Room 3/4* 1 - Application of NGS to improving diagnostic yield for Infantile-onset epileptic encephalopathies *Elizabeth Palmer* 2 - Comparative heath economic costings of next generation sequencing versus traditional neurometabolic investigation in a cohort of children with epileptic encephalopathy *Rani Sachdev* 3 - Next generation sequencing provides answers for families affected by foetal akinesia, arthrogryposis, and severe congenital myopathies *Emily Todd*	**CONCURRENT 2: ASGC** *Chair: Alison McEwen and Mary-Anne Young* *Ballroom C* 1 - Psychological impact of inconclusive BRCA1/ test results *Jennifer Berkman* 2 -Case study of a long standing support group for Jewish *BRCA* mutation carriers *Suzannah Bawden* 3 -Preconception Genetic Screening: Reflections on the first 26 patients to undergo the Counsyl test in a private genetic counselling setting *Ron Fleischer* 4 - Exploring the psychosocial support services for women with divergence of sex development *Chloe Hanna*