HGSA SPECIAL INTEREST GROUP MEETINGS – 17-18 NOVEMBER 2023 – MELBOURNE VIC

FRIDAY 17 NOVEMBER 2023

SIG	ASIEM NEWBORN SCREENING	ASIEM TRAINEES / ALLIED HEALTH		ASDG	AACG	ASGC
Room	**MEETING PLACE 1**	**MEETING PLACE 2**		**MEETING PLACE 3**	**MEETING PLACE 4**	**CENTRE STAGE 2**
0830-1030	**SESSION 1** **CLOSED SESSIONS**		**0830-1000**	**SESSION 1**		
	Newborn screening Chair: Dianne Webster 0830: NZ update 0840: NSW update 0850: SA update 0910: Vic update 0920: WA update 0930: National (NZ) and federal (Aus) Policy implementation 1000: Discussion and Q&A	**ASIEM Academy** Facilitated by Carolyn Bursle and Kalliope Demetriou		**Welcome** **Submitted Orals - Learning from each other to improve patient care** 1. DEVELOPING A PROCESS FOR BIOINFORMATIC IMPACT ASSESSMENT OF CIS-REGULATORY VARIANTS IN MONOGENIC DISEASE - Dr Rehan Villani 2. A TIME TO REVERT: A SEAMLESS CLINICAL, RESEARCH AND DIAGNOSTIC APPROACH TO ACHIEVING A DIAGNOSIS - Dr Ee Ming Wong *3.* PILOTING AN INTERPRETIVE MODULE FOR GENOMIC TESTING FOR CHILDHOOD SYNDROMES AND INTELLECTUAL DISABILITY – Prof Bruce Bennetts 4. OCULAR GENOMIC MULTIDISCIPLINARY TEAM (OCULARGEN-MDT): A NATIONAL RESOURCE FACILITATING MOLECULAR DIAGNOSES AND THERAPY ELIGIBILITY IN THE INHERITED RETINAL DYSTROPHIES - Dr Benjamin Nash 5. ASDG CONVENTIONAL CHROMOSOMAL MICROARRAY ANALYSIS BEST PRACTICE GUIDELINES - CONSIDERATIONS FOR REPORTING OF GERMLINE UNSOLICITED FINDINGS FROM THE WORKING PARTY – Miss Amber Burgess	**Dysmorphology cases** CLOSED SESSION	**Welcome –** Ms Lisette Curnow and A/Prof Laura Forrest **Interactive education session: Intersectionality and Trans-Inclusive Care in Genomic Medicine** Chairs: Rebecca Purvis, Joshua Schultz, Anastasia Lewis, and Lucas Mitchell (virtual) International speakers (virtual): Prof Jehannine Austin and A/Prof Kim Zayhowski
			1000-1030	**MORNING TEA & DISPLAYS**		

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<tr><td>1030-1100</td><td colspan="2">MORNING TEA & DISPLAYS</td><td>1030-1200</td><td colspan="3">SESSION 2</td></tr>
<tr><td>1100-1245</td><td colspan="2">SESSION 2
CLOSED SESSIONS</td><td></td><td rowspan="2">1030-1115
Invited Speaker Presentation:
The old and the new: A review of current cytogenetic and genomic testing methodologies for haematological malignancies at Pathology Queensland
Dr Adayapalam Nandini

1115-1200
MAVE Education Group session</td><td rowspan="2">1030-1115
Invited Speaker Presentation:
The role of Health Economics in the evaluation and translation of Genomic Medicine
A/Prof Ilias Goranitis

1115-1200
AACG AGM
CLOSED SESSION</td><td rowspan="2">1030-1100
Chair: Dr Marta Cifuentes Ochoa and Mr Ivan Macciocca
Invited speaker Presentation:
The value of ancestry specific genomic data for genetic counselling practice
Ms Yasmin Blystra

1100-1200
Submitted Oral Presentations

1. PSYCHOSOCIAL ISSUES RAISED DURING GENETIC COUNSELLING FOR INDIVIDUALS AND FAMILIES OF MIXED ETHNICITY AND CULTURE: EXPLORING AUSTRALIAN GENETIC COUNSELLORS' PERSPECTIVES – Mr Parker Truong
2. IT'S ABOUT US: A QUALITATIVE EXPLORATION OF SOUTH ASIAN COMMUNITY ATTITUDES TOWARDS GENOMIC RESEARCH – Ms Vaishnavi Nathan
3. DEFINING NEXT STEPS IN THE IMPLEMENTATION OF POLYGENIC SCORES IN AUSTRALIA'S GENETICS CLINICS: PROFESSIONALS' VIEWS ON DETERMINANTS, STRATEGIES AND NEEDS – Ms Rebecca Purvis
4. A QUALITATIVE EXPLORATION OF GENETIC HEALTHCARE PROVIDERS LEARNING NEEDS AND PREFERENCES FOR POLYGENIC RISK SCORE EDUCATION - Mrs Amy Clark</td></tr>
<tr><td rowspan="2"></td><td colspan="2" rowspan="2">1100-1245

Newborn screening research meeting
Chair: Natasha Heather

1100: Prof Bruce Bennetts - NEWBORN GEN SEQ TRAIL

11.15: A/Prof David GODLER -EpiGNs

1130: A/Prof Karin Kassahn - Newborns in SA

1145: A/Prof Sarah Norris - gEnomics4newborns

1200; A/Prof Sebastian Lunke - BabyScreen+

1215: Discussion</td><td rowspan="2">1100-1300

ASIEM Academy
Facilitated by Carolyn Bursle and Kalliope Demetriou</td><td></td></tr>
<tr><td>1200-1300</td><td colspan="3">LUNCH, DISPLAYS & POSTERS</td></tr>
<tr><td>1245-1330</td><td colspan="2">LUNCH, DISPLAYS & POSTERS</td><td>1300-1430</td><td></td><td colspan="2">SESSION 3</td></tr>
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				1300-1345 **ASDG AGM** CLOSED SESSION		
1330-1430	**SESSION 3** **CLOSED SESSIONS**				**Dysmorphology cases** **CLOSED SESSION**	**CLOSED SESSION** **Family Therapy workshop: Becoming unstuck – the use of Family therapy techniques in Genetic Counselling** Facilitators: Nicky Gelfand, Tiffany O'Brien, Lisa Gordon, and Anthony Hurst **Interesting cases** 1. THE COMPLEXITIES OF NAVIGATING MULTIPLE FAMILY MEMBERS ACROSS GENERATIONS-WHO IS OUR PATIENT? – Ms Lisa Gordon 2. COMPLEX FAMILY DYNAMICS IN A SIBSHIP AT RISK OF HUNTINGTON DISEASE – Ms Jennifer Berkman
	Newborn screening (Newborn Screening Committee) **Harmonisation of Newborn screening and collection**	**Metabolic Genetics (submitted orals)** 1. PREGNANCY WEIGHT GAIN IN MATERNAL PHENYLKETOURIA (MPKU) – Ms Amy Allia 2. A DARS2 MILD LEUKODYSTROPHY AND CHALLENGES INTERPRETING AN INTRONIC SPLICING HYPOMORPH AS SECOND HIT – Dr Michel Tchan 3. DETAILED NEUROPSYCHOLOGICAL EVALUATIONS IN ATTENUATED METACHROMATIC LEUKODYSTROPHY – Dr Michel Tchan 4. RETROSPECTIVE DIAGNOSIS OF OTC DEFICIENCY IN A DECEASED NEONATE FOLLOWING PRESENTATION IN A SUBSEQUENT SIBLING – Dr Sarah Donoghue		**1345-1430** **SESSION 3** **Submitted Orals - Genomics** 1. MOSAIC SEGMENTAL OVERGROWTH SYNDROMES GENE SEQUENCING ON THE ION TORRENT NGS PLATFORM - Mr George Elakis 2. SOMATIC GENOMIC TESTING CAPACITY AND CURATION PRACTICES IN AUSTRALIAN AND NEW ZEALAND DIAGNOSTIC TESTING LABORATORIES - Ms Grace Pendlebury **3.** TECHNICAL LIMITATIONS OF PKD1 GENE SEQUENCING - VCGS EXPERIENCE - Dr Suliman Khan		
1430-1500	**AFTERNOON TEA, DISPLAYS, POSTERS**					
1500-1600	**HGSA EGM**					

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1600-1730	**SESSION 4**					
	1600-1640 **Wilcken / Danks Lecture:** **Solving puzzles in biochemical genetics** Dr James Pitt **1640-1730** **ASIEM AGM**			**1600-1630** **QAP** **1630-1700** **Submitted Orals - Panel testing** 1. THE "GOLDILOCKS" PANEL: DETERMINING THE OPTIMAL NUMBER OF GENES TO INCLUDE WHEN PERFORMING REPRODUCTIVE CARRIER SCREENING - Ms Mia Gruzin 2. IMPLEMENTATION OF NEW CARDIAC PANEL ANALYSIS SERVICE AT NSWHP RANDWICK GENOMICS - Ms Melissa Cullen **1700-1730** **ASDG Mentoring Program Launch**	**Dysmorphology cases** CLOSED SESSION	**1600-1700** Chairs: Ms Anna Jarmolowicz and Ms Lisette Curnow **Submitted Oral Presentations** - CARERS LIVED EXPERIENCE OF FAMILIAL ID AND THEIR ATTITUDES TOWARDS REPRODUCTIVE TECHNOLOGY – Miss Hannah Thomson - THE IMPACT OF GENETIC COUNSELLING ON PEOPLE WITH AN INCREASED-CHANCE REPRODUCTIVE GENETIC CARRIER SCREENING RESULT: A MIXED-METHODS STUDY - Miss Ella Zurita - IVF SUCCESS RATES IN INDIVIDUALS ACCESSING PREIMPLANTATION GENETIC TESTING FOR MONOGENIC CONDITIONS (PGT-M) IN VICTORIA, AUSTRALIA – Ms Alice Poulton - "MORE THAN JUST A TEST" - A QUALITATIVE STUDY ON THE CLINICAL UTILITY OF GENOMICS IN RARE DISEASES – Mr Ryan Pysar (virtual) **1700-1730** Chair: Ms Lisette Curnow **Judith Elber Presentation** **Dr Samantha Wake**
1730-2030	**SIG MIXER – The Terrace, Rydges Melbourne**					

SATURDAY 18 NOVEMBER 2023

SIG	ASIEM	ASDG	AACG	ASGC
ROOM	MEETING PLACE 1	MEETING PLACE 2&3	MEETING PLACE 4	CENTRE STAGE 2
0900-1030	SESSION 5			
	Biochemical Genetics Chair James Pitt **0900-0930** **Invited Speaker Presentation speaker** **RDMassSpec: Ultra rapid proteomics for rare disease** A/Prof David Stroud **0930-1030** **Submitted Orals** 1. THERAPEUTICS FOR NAXD DISORDER – A NEWLY DESCRIBED FATAL ENCEPHALOPATHY TRIGGERED BY FEVER – Dr Nicole Van Bergen 2. STEM CELL MODELLING OF MITOCHONDRIAL DISEASE-LINKED CARDIOMYOPATHY – Dr Ann Frazier 3. PROLIDASE DEFICIENCY: A CASE REPORT AND IMPROVED SPECIFICITY OF URINE SCREENING – Miss Abisha Srikumar 4. DETECTION OF A SEVERE FORM OF ADENYLOSUCCINATE LYASE DEFICIENCY BY FIRST TIER URINE METABOLIC SCREENING WITH TARGETED TANDEM MASS SPECTROMETRY – Mr Kai Mun Hong	**0900-1000** **Invited Speaker Presentation** **Insights into the ACMG/AMP/CAP/ ClinGen Standards for Sequence Variant Classification v4.0** Dr Alicia Byrne **1000-1030** **Submitted Orals – Cytogenetics** 1. GENOMIC CHARACTERISATION OF GATA2 DEFICIENCY SYNDROME. A CASE REPORT - Mrs Naomi Westland 2. TWO CASES OF MOSAIC RING CHROMOSOME 22 WITH PHELAN-MCDERMID SYNDROME ASSOCIATED WITH INCREASED TUMOUR RISK - Miss Bianca Akhurst	**0900-0945** **Submitted Orals** 1. MAKING SENSE OF THE COMPLEX: LONG READ SEQUENCING DELINEATING COMPLEX STRUCTURAL VARIANTS IN RARE GENETIC CONDITIONS - Dr Lisa Ewans 2. THE PSYCHOSOCIAL IMPACT AND HEALTH OUTCOMES OF GENETICS CONSULTATIONS AND TESTING IN AUSTRALIA: THE ENTRUST STUDY - A/Prof Kathy Wu 3. GENOTYPING IN PAEDIATRIC VASCULAR ANOMALIES: THE IMPACT OF MOLECULAR DIAGNOSIS ON CLINICAL PRACTICE - Dr Sinead O'Sullivan **0945-1030** **Invited Speaker Presentation** **Vascular malformations research** Dr Natasha Brown	**0900-0930** Chair: Ms Giulia Valente and Dr Helen Mountain **Invited Speaker Presentation:** **A genetic counselling led, adaptive, translation program returning genomic research results to research participants** Ms Mary-Anne Young **0930-1030** **Submitted Oral Presentations** 1. HOW TO MANAGE 'INCREASED CHANCE' RESULTS FROM GENOMIC NEWBORN SCREENING: FINDINGS FROM KEY INFORMANT INTERVIEWS – Ms Erin Tutty 2. BECOMING AGENTS FOR GENOMIC CHANGE: GENETIC COUNSELLORS' VIEWS OF MAINSTREAMING GENOMICS IN PATIENT CARE AND IMPLEMENTATION INFLUENCES - Trang Do 3. EVALUATING CO-DESIGNED INTERVENTIONS SUPPORTING PAEDIATRICIANS TO ORDER FUNDED GENOMIC TESTS: A WEBSITE, 'ASK-A-GENETICS-EXPERT' CONTACT SERVICE AND AWARENESS RAISING ACTIVITIES – A/Prof Belinda Dawson-McClaren 4. WHAT MATTERS TO PARENTS? BUILDING THE FOUNDATIONS OF A PARENT-REPORTED EXPERIENCE MEASURE FOR THE MAINSTREAM DELIVERY OF GENOMIC TESTING – Ms Erin Crellin
1030-1100	MORNING TEA, DISPLAYS & POSTERS			

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<tr><td>1100-1230</td><td colspan="4">SESSION 6</td></tr>
<tr><td></td><td>Newborn Screening – Submitted Orals
Chair Natasha Heather

1. INTRODUCTION OF SUCCINYLACETONE AS A NEWBORN SCREENING MARKER FOR TYROSINAEMIA – THE NEW ZEALAND EXPERIENCE – Mr Mark De Hora
2. MONITORING PHENYLALANINE IN A NEWBORN SCREENED INFANT WITH TYROSINAEMIA TYPE 1 – Mrs Rychelle Winstone
3. HEREDITARY TYROSINEMIA TYPE 1 IN MELBOURNE, AUSTRALIA: A 22-YEAR RETROSPECTIVE STUDY – Dr Dinusha Pandithan
4. TRANSIENT INCREASED 11-DEOXYCORTISOL DETECTED ON NEWBORN BLOODSPOT SCREENING FOR CONGENITAL ADRENAL HYPERPLASIA – Mr Lawrence Greed
5. MANAGING NEWBORN SCREENING RECOLLECTIONS FOR SICK AND PRETERM NEONATES – A/Prof Ronda Greaves

Session Q & A</td><td colspan="2">1100-1145 (MP 2&3)
Getting Stuff done: a guide for the distracted academic.
Prof Inger Mewburn

1145-1220
Poster Prize presentations

1. KANSL1 GENOTYPING COMPLEX REGIONS AND COPYNUMBERS - Mr Luke Wainwright
2. MOSAIC DCX VARIANTS IN PATIENTS WITH SUBCORTICAL BAND HETEROTOPIA - Mr Kristian Brion
3. EIGHT SEQUENTIAL FISH ROUNDS ON BLOOD SMEARS IDENTIFIED ETV6::MNX1 ASSOCIATED WITH HIGH-RISK CYTOGENETICS IN INFANT ACUTE MYELOID LEUKAEMIA - Ms Christina Chou
4. UNEXPECTED IDENTIFICATION OF A CONTIGUOUS GENE DELETION BY REPRODUCTIVE GENETIC CARRIER SCREENING - Ms Crystle Lee
5. NOT SO MUTUALLY EXCLUSIVE: A DOUBLE DRIVER MPN CASE STUDY - Mrs Tamaryn Knezovich (virtual)
6. TOO MANY SNPS SPOIL THE BROTH: OTC PROBE LOSS IN SNP MICROARRAY TESTING - Mr Oliver Van Wageningen (virtual)</td><td>1145-1230
Submitted Orals

1. CLINICAL AND GENOMIC TESTING OUTCOMES IN CHILDREN WITH SEVERE OR RECURRENT HYPOGLYCAEMIA AT THE ROYAL CHILDREN'S HOSPITAL 2016-2021 - Dr Oliver Heath
2. BLOOD-BASED BIOMARKER FOR SPAST-ASSOCIATED HEREDITARY SPASTIC PARPALEGIA (HSP-SPAST) - Dr Sue-Faye Siow
3. 7 NOVEL VARIANTS OF WEISS-KRUSZKA SYNDROME IN THE AIM TO EXPAND PHENOTYPE - Dr Anna Hau (virtual)</td><td>1100-1230
CLOSED SESSION
Ethics workshop including interesting cases
Facilitators: Ms Sharon Feldman & Dr Melody Menezes

Interesting Cases

1. WHOSE RIGHT IS IT ANYWAY? A REQUEST FOR PRE-SYMPTOMATIC HD TESTING IN A CHILD WITH LIMITED CAPACITY – Ms Suzannah Bawden
2. THE ETHICAL IMPACTS OF PROCEDURAL CHANGES ON PATIENTS AND THEIR FAMILIES- Ms Laura Barth
3. GENETIC TESTING FOR BREAST CANCER IN A TRANSGENDER PATIENT - Dr Gabrielle Reid</td></tr>
<tr><td>1230-1330</td><td>LUNCH, DISPLAYS, POSTERS</td><td>1230-1400</td><td colspan="2">LUNCH, DISPLAYS & POSTER SESSION (1300-1330)</td></tr>
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1330-1530	**SESSION 7**			
	1330-1400 **Invited Speaker Presentation** **Exercise testing and prescription in patients with inborn errors of muscle metabolism** Ms Kiera Batten	**1400-1530**	**SESSION 7**	
	1400-1530 **Submitted Orals** 1. EFFECTS OF COMMENCING SAPROPTERIN THERAPY ON QUALITY OF LIFE FOR CHILDREN WITH PHENYLKETONURIA AND THEIR FAMILIES: A QUALITATIVE INTERVIEW STUDY – Ms Catherine Atthow 2. DIETARY MANAGEMENT OF HMG-COA LYASE DEFICIENCY – SIMPLIFYING THE COMPLEXITY – Mrs Susan Thompson 3. COMPARISON OF MPKU DIETARY PROTEIN INTAKE BEFORE AND AFTER TETRABIOPTERIN (BH4) – Mrs Clare Williams 4. MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOMYOPATHY: WATCH THE THYMINE! – Dr Rebecca Quinn 5. A NOVEL MUTATION IN CPS-1 GENE RESULTING IN LATE ONSET CPS-1 DEFICIENCY – Dr Suchitra Raj 6. CRITICALLY UNWELL CHILDREN WITH MITOCHONDRIAL DISORDERS DIAGNOSED BY ULTRA-RAPID GENOMIC SEQUENCING – Dr Megan Ball	**1400-1530** **ASDG Cytogenetics Best Practice Guidelines Drafts** Presentation and discussion	**1400-1530** **Submitted Orals** 1. CLINICAL OUTCOMES FOLLOWING PRE-IMPLANTATION GENETIC TESTING FOR MONOGENIC CONDITIONS, A SYSTEMATIC REVIEW - Ms Alice Poulton 2. PRENATAL WHOLE EXOME SEQUENCING: DIAGNOSES AND DILEMMAS - Mr Ron Fleischer & Miss Bethany Wadling 3. PREGNANCY AND BIRTH OUTCOMES FOLLOWING MOSAIC EMBRYO TRANSFERS - Miss Tamara Mossfield 4. GENE SELECTION FOR GENOMIC NEWBORN SCREENING: MOVING TOWARDS CONSENSUS? - Dr Lilian Downie 5. SUPPORTING REPRODUCTIVE DECISION MAKING IN NEUROFIBROMATOSIS (NF-SURE) - Dr Alison McLean **6.** NEUROFIBROMATOSIS MODEL OF CARE PROJECT: DEVELOPMENT OF A STATE-WIDE INTEGRATED VALUE-BASED MODEL OF CARE FOR THE NEUROFIBROMATOSES - Dr Sue-Faye Siow	**1400-1500** CLOSED SESSION Chair: Ms Sachini Poogoda and Ms Elly Lynch **Interesting cases** 1. GENETIC COUNSELLING CHALLENGES IN INTERPRETING TP53 PATHOGENIC VARIANTS IN GERMLINE GENETIC TESTING FOR BREAST CANCER PATIENTS WITH POSSIBLE MOSAIC RESULTS - Ms Sook-Yee Yoon 2. SHAME AND GUILT IMPACTING ON THE DISCLOSURE OF CONSANGUINITY - ASGC INTERESTING CASE - Ms Kate Riley 3. ULTRA-RAPID WHOLE GENOME SEQUENCING AT 38 WEEKS GESTATION FOR VENTRICULOMEGALY AND ANTERIOR TEMPORAL POLE CYSTS – Ms Helen Kincaid 4. AN INCIDENTAL FINDING OF A TP53 PATHOGENIC VARIANT IN THE PRENATAL SETTING - Yael Prawer Interesting Cases Q&A **1500-1530** Chair: Ms Laura Barth and Ms Elly Lynch **Invited Speaker Presentation:** **Well-being strategies for cardiac genetic counsellors** Ms Laura Yeates

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1530-1600	**AFTERNOON TEA**	**CLOSING & AWARD PRESENTATIONS**	**CLOSING & AWARD PRESENTATIONS**	**CLOSING & AWARD PRESENTATIONS**
1600-1700	**SESSION 8**			
	Metabolic Genetics **1600-1630** **Oral Sepiapterin in Phenylketonuria: Results from the phase 3 APHENITY trial and open label, long-term extension study** Dr Michel Tchan **1630-1700** **Submitted Orals** 1. POTENTIAL DISEASE MODIFYING EFFECTS AND CLINICAL OUTCOMES IN SUBJECTS WITH MUCOPOLYSACCHARIDOSIS I TREATED WITH PENTOSAN POLYSULFATE SODIUM - Dr David Ketteridge 2. SERTRALINE USE IN PATIENTS WITH GLUTARIC ACIDURIA TYPE 2 ON ACYLCARNITINE PROFILE AND LACK OF PATHOGENIC BIALLELIC VARIANTS IN ETFA/ETFB/ETFDH – Dr Liza Phillips **Close of Meeting**			