Programme

SATURDAY 3 AUGUST 2019	
0730-1800	**REGISTRATION DESK** *Fletcher Challenge Floyer, Grd Fl*
0730-1400	**HGSA Council Meeting** Wharewaka Function Centre, Matiu Room
0815-1300	**MHGSA Exams** Michael Fowler Centre, VIP Room, L2
	SPECIAL INTEREST GROUP MEETINGS Michael Fowler Centre

HGSA 43RD ANNUAL SCIENTIFIC MEETING FORMALLY OPENS	
1700-1720	**WELLINGTON COLLEGE & WELLINGTON GIRLS COLLEGE** **TE HAEATA AWATEA - KAPA HAKA** **AUDITORIUM, L1**
1720-1915	**Welcome Reception & Exhibition Opening** – Renouf Foyer & Upper Promenade, L1 *Join delegates, sponsors and exhibitors for the first networking function of the meeting.* *This is the official opening of the Exhibition.*
1930-2200	**AACG Dinner** **Noble Rot Wine Bar** (6 Swan Ln, Te Aro, Wellington) **(For those who have pre-booked only)**
1930-LATE	**ASDG SIG Mixer** **St Johns Bar** (5 Cable St, Wellington)
1930-2300	**ASGC Mixer** **Waterfront Room, Harbourside Function Centre** (4 Taranaki St Wharf, Wellington)
1930-2130	**ASIEM SIG Mixer** **The Library** (Upstairs, 53 Courtenay Place, Wellington)

SUNDAY 4 AUGUST 2019	
0730-1800	**REGISTRATION DESK** – *Fletcher Challenge Foyer, Grd Fl*
0830-0845	**OPENING CEREMONY** – *Auditorium, L1*
0845-1015	**OPENING PLENARY & THEMED SESSION 1: POPULATION GENOMICS** – *Auditorium, L1* *Chairs: Dr David Amor & Dr Dianne Webster* Plenary 1: Population genomics and the UK 100,000 Genomes Project experience ***– Dr Richard Scott*** Plenary 2: The Aotearoa New Zealand Genomic Variome Project ***– Dr Stephen Robertson*** Plenary 3: Mackenzie's Mission ***– Prof Martin Delatycki, Prof Nigel Laing, Prof Edwin Kirk*** Plenary 4: The ethical implementation of genomics in population health ***– Dr Ainsley Newson***
1015-1045	**MORNING TEA & EXHIBITION** – *Renouf Foyer & Lower Promenade, L1*
1045-1215	**PLENARY & THEMED SESSION 2: INTRODUCTION OF GENOMICS INTO CLINICAL CARE** – *Auditorium, L1* *Chairs: Dr Kate Neas & Dr Richard King* Plenary 5: Future of Genomic Medicine ***– Dr Wendy Chung*** Plenary 6: Delivery of ethical and effective genomic health care ***– Dr Christine Patch*** Plenary 7: It doesn't just happen: Changing Healthcare ***– A/Prof Clara Gaff*** Submitted Oral (part of Top 15 Orals): From '9 to 5' to '24/7': Delivering rapid genomics at scale in a diagnostic laboratory ***– Dr Sebastian Lunke***
1215-1315	**LUNCH, POSTERS & EXHIBITION** *Renouf Foyer & Lower Promenade, L1 & Fletcher Challenge Floyer, Grd Fl*

SUNDAY 4 AUGUST 2019	
1315-1445	**CONCURRENT SIG SESSIONS**
	CONCURRENT SESSION 1: AACG – *Frank Taplin Room, L1* *Chair: A/Prof Paul James* The diagnostic trajectory of families undiagnosed after singleton exome sequencing ***– Mr Thomas Cloney*** What to do with a negative exome? ***– Dr Natalie B. Tan*** Vesicular GABA co-transporter variants are associated with genetic epilepsy with febrile seizures plus (GEFS+) ***– Dr Sarah Heron*** Exploration of interactions between genetic health professionals and laboratory specialists in clinical genomic sequencing ***– Dr Danya Vears*** Genetics of functional bowel disorders: looking for genes where the sun don't shine ***– Prof Mauro D'Amato*** Population Genetic Testing for Breast and Ovarian Cancer Susceptibility ***– A/Prof Alison Trainer***
	CONCURRENT SESSION 2: ASGC – *Auditorium, L1* *Chairs: Dr Amy Nisselle & Mrs Subhashini Crerar* Genetic testing for familial motor neurone disease (MND): insights and challenges ***– Ms Ashley Crook*** Expanded genetic carrier screening – lessons learnt from the first 130 couples ***– Ms Zoe Milgrom*** General Practitioners and genomics: their views on practice and education in Australia ***– Ms Marie Cusack*** Ethical implications of population–scale genomic screening ***– Ms Jane Tiller*** Acceptability and feasibility of a pre–clinic psychosocial screening tool in an Australian clinical genetics setting ***– Miss Rebecca Purvis*** How do adolescents and young adults experience genetic testing for Li-Fraumeni syndrome? ***– Mr Rowan Forbes Shepherd***
	CONCURRENT SESSION 3: ASIEM – *Lion Harbour View 1, L2* *Chairs: A/Prof Carolyn Ellaway & A/Prof Ronda Greaves* Bile acid disorders ***– Prof Peter Clayton*** Extreme phenotype sampling and whole genome sequencing of Niemann–pick type C disease in Australasia ***– Dr Andrew Munkacsi*** Outside The Exome: The role of CNVs, SVs, deep-intronic and 'silent' mutations in Mitochondrial disease ***– Dr Alison G Compton*** Mitochondrial Disease in New Zealand: A nationwide prevalence study ***– Dr Sarah Missen***

SUNDAY 4 AUGUST 2019

CONCURRENT SESSION 4: ASDG – *Lion Harbour View 2, L2*
Chair: Mr Ben Lundie

A Customisable Analysis Pipeline for Identification of Clinically Relevant Genetic Variants in Next Generation Sequencing Data
– Dr Miles Benton

Detecting Repeat Expansions with sequencing data: implications for novel discoveries and rapid genetic diagnosis ***– Prof Melanie Bahlo***

Whole genome sequencing reveals structural gene rearrangements and novel splicing variants in the retinal dystrophies
– Mr Benjamin Nash

Genomics education in the UK: what can we learn?
– Mrs Jo Martindale

Quality Control is critical for Genomic Analysis ***– Ms Nila Quayum***

1445-1515 **AFTERNOON TEA & EXHIBITION** – *Renouf Foyer & Lower Promenade, L1*

1515-1615 **CONCURRENT INDUSTRY SESSIONS**

Illumina Industry Session *Auditorium, L1*

illumina®

Whole Genome Sequencing in Clinical Practice
Dr John Belmont
Sr. Principal Medical Scientist, Illumina Inc.

PerkinElmer Industry Session *Lion Harbour View 1, L2*

PerkinElmer®
For the Better

The Future of Genomics in Clinical Screening
Introduction
Screening for neuromuscular disorders and immunodeficiencies
A/Prof Veronica Wiley
Clinical Associate Professor Genetic Medicine, Children's Hospital, Westmead NSW Australia

GenOMICS for Precision Medicine
Mr Ephrem Chin
Director, Global Lab Operations PerkinElmer Genomics

Q&A

SUNDAY 4 AUGUST 2019

Agilent Technologies Industry Session *Lion Harbour View 2, L2*

Agilent

Target Next Generation Sequencing – Enabling Precision Medicine

Exomes at VCGS: A look under the hood
Dr Stefanie Eggers
PhD, Head of Sequencing Service Platform and co–lead of VCGS Development Team (Assays and Technologies)

Victorian Clinical Genetics Services Agilent's NGS solutions: Addressing Today's Challenges and Enabling Precision Medicine
Dr Dan Belluoccio
Sr Field Application Scientist, Agilent Technologies

Q&A Session

CONCURRENT WORKSHOPS

1515-1815	**RACP SPDP Workshop** – *Frank Taplin Room, L1*
1615-1815	**Variant Curation Workshop (Introductory)** – *Auditorium L1* This workshop is aimed at those who would like a gentle introduction to the ACMG variant curation guidelines and some of the tools that can be used. The plan for this workshop is to work through some variants in a guided way, using your own devices.
	Variant Curation Workshop (Advanced) – *Lion Harbour View 1* This workshop is for those who have more confidence in their curation skills and will have an expert panel of curators who will lead discussion of some variants submitted by workshop attendees.
	Genomics Education Network of Australasia Workshop – *Civic Café, Grd Fl*

SUNDAY 4 AUGUST 2019

1615-1815	**ASIEM Dietetic Workshop** *Lion Harbour View 2, L2*
	Chairs: Rhonda Akroyd and Rychelle Winstone
	**Ms Rachel Skeath (invited speaker) to offer comment and support discussion*
	ASIEM Oral Presentation:
1615	KD in McArdle disease : a case study **– *Rychelle Winstone***
1630	Discussion KD in IEM
	Short Cases
1640	Newly Diagnosed adult MADD/GAII with Guillain-Barré syndrome **– *Clare Williams***
1645	Discussion
1655	Low BCAA in Argininosuccinic acidaemia **– *Sue Thompson***
1700	Discussion
1710	GSD1a: Prioritising and preserving normal feeding development **– *Ciara Paramore***
1715	Discussion
1725	Supporting PKU women, reflections on inaugural PKU Mums morning tea **– *Anne-Marie Desai***
1730	Discussion
	Discussion: Challenges Facing Metabolic Dietitians
1740	Taking IEM patient/carer teaching into the 21st century **– *Rychelle Winstone***
1745	Discussion
1755	Blended tube diet in IEM **– *Rhonda Akroyd***
1800	Discussion
1810	**Summary and Close**

1815-1915	**Posters Session** Includes platters & drinks. Posters presenters will be available at their poster during the following times. 1815–1845 – Odd numbered posters 1845–1915 – Even numbered posters *Renouf Foyer & Lower Promenade, L1 & Fletcher Challenge Foyer, Grd Fl*	**1815–1845** University of Technology Sydney and University of Melbourne 'Master of Genetic Counselling' Information Session *Lion Harbour View 1, L2*

MONDAY 5 AUGUST 2019	
0700-0820	**Sanofi Genzyme Industry Session: Newborn screening and the management of late-onset patients** – *Auditorium, L1* *Includes a light stand-up breakfast in the foyer from 7am.* SANOFI GENZYME **Welcome and Introduction** – *Dianne Webster (Chair)* *Director Newborn Metabolic Screening Programme, Auckland District Health Board* **Current state of play? An overview of NBS expansion around the world – *Bridget Wilcken*** *Senior Staff Specialist, Centre for Clinical Genetics, Sydney Children's Hospital* **Developing technologies in NBS – how can we separate early vs late disease vs false-positives? – *Veronica Wiley*** *Principal Scientist/ Director – NSW Newborn Screening Program* **What is available vs what is needed for long-term follow up of late onset patients (i.e. a gap analysis of current practices). – *Drago Bratkovic*** *Head, Metabolic Unit – Women's and Children's Hospital, North Adelaide* Q & A
0730-1700	**REGISTRATION DESK** - *Fletcher Challenge Foyer, Grd Fl*
0830-1000	**CONCURRENT SIG SESSIONS**
	Concurrent Session 5: AACG – *Frank Taplin Room, L1* *Chairs: Dr Melanie Zeppel & Prof Stephen Robertson* Human Medical Genetics and Genomics Competencies for the contemporary Medical School Graduate **– *A/Prof Diane Kenwright*** The Ethics of NIPT: Women's Experiences and Attitudes on Expanding the Reach **– *Ms Hilary Bowman-Smart, Prof Julian Savulescu*** Reproductive preferences in parents with lived experience of caring for their children with intellectual disability **– *Ms Radhika Rajkumar*** Preimplantation Genetic Diagnosis (PGD) for retinoblastoma survivors: cost-effectiveness and quality of life improvements **– *Dr Melanie Zeppel*** Comparison between preconception carrier screening for spinal muscular atrophy and treating it with nusinersen: a cost-effectiveness analysis **– *Dr Rupendra Shrestha*** Maximising the Minimum Gain: Ensuring Equity of Access to BRCA Testing **– *A/Prof Alison Trainer***

MONDAY 5 AUGUST 2019	
	Concurrent Session 6: ASGC – *Auditorium, L1* *Chairs: Ms Emma Felix & Prof Christine Patch* Taking a Family Health History: What shapes the Aboriginal and Torres Strait Islander story? **– Dr Mona Saleh** Developing guidelines for genomic researchers partnering with Aboriginal and Torres Strait Islander people of Queensland **– *Dr Miranda Vidgen*** Parent experiences with ultra–rapid genomic sequencing in paediatric acute care **– *Ms Gemma Brett*** Ethical Challenges in Genomic Testing in the NICU: Clinicians' views **– *Dr Todor Arsov*** Genetic counsellors in the NICU and PICU: Experiences from the Australian Acute Care Genomics project **– *Dr Michelle G. de Silva*** Clinical leadership and genomics – what works? **– *Dr Stephanie Best***
	Concurrent Session 7: ASIEM – *Lion Harbour View 1, L2* *Chairs: A/Prof Veronica Wiley & Dr Natasha Heather* The confluence of Genomic Sequencing and Newborn Screening **– *Prof Cynthia Powell*** Growing up with a hidden disorder: An ethnography of the metabolic condition MCADD in New Zealand **– *Dr Pauline Herbst*** Baby Beyond Hearing, using genomics as a newborn screening tool **– *Dr Lilian Downie*** Newborn Screening of Severe Combined Immune Deficiency in NZ **– *Dr Pippa Dryland***
0830-0900	**Prenatal Array Workshop – *Ms Rachel Beddow*** – *Lion Harbour View 2, L2*
0900-1000	**Concurrent Session 8: ASDG** – *Lion Harbour View 2, L2* *Chair: Dr Amanda Dixon–McIver* Identification and characterisation of predisposition genes and mutations in familial haematological and pan-cancer families **– *Ms Julia Dobbins*** A Clinic Based Multidisciplinary Review for Variants of Uncertain Significance identified in Cancer Susceptibility Genes **– *Ms Alexandra Lewis*** Somatic mutation testing for cancer services In Queensland public hospitals **– *Dr David Fairbairn*** Whole genome sequence analysis of patients with suspected hereditary cancer **– *Ms Aimee Davidson***

MONDAY 5 AUGUST 2019	
1000-1045	**MORNING TEA & EXHIBITION** - *Renouf Foyer & Lower Promenade, L1*
1045-1200	**Plenary & Themed Session 3: Cancer** – *Auditorium, L1* *Chairs: A/Prof Alison Trainer & Ms Vanessa Tyrrell* Plenary 8: Mainstreaming in cancer genetics **– *A/Prof Kathy Tucker*** Plenary 9: Chimeric Antigen Receptor T-Cells: A Cell and Gene Therapy for Cancer **– *Dr Robert Weinkove*** Plenary 10: CAPP3 and risk management of familial Colorectal Cancer **– *Dr Finlay Macrae***
1045-1200	**ASIEM Allied Health & Nurses Session** – *Frank Taplin Room, L1* *Chairs: Miss Rebecca Nicol & Mrs Sue Thompson* Dietetic Challenges in managing Glycogen Storage Diseases (GSD): Focusing on GSD I and III **– *Ms Rachel Skeath*** Continuous glucose monitoring in hepatic glycogen storage disorders: A systematic review **– *Ms Kristen Fitzell*** The use of Tetrahydrobiopterin in PKU: the Victorian Experience **– *Dr Maureen Evans***
1200-1245	**Sutherland Lecture** – *Auditorium, L1* *Chairs: A/Prof Alison Trainer & Ms Vanessa Tyrrell* Translating complex genetics into practice: The example of breast cancer risk **– A/Prof Paul James**
1245-1415	**LUNCH, POSTERS & EXHIBITION** Includes concurrent Poster Sessions (Top posters) *Renouf Foyer & Lower Promenade, L1 & Fletcher Challenge Foyer, Grd Fl*
1330-1400	**CONCURRENT SESSIONS – TOP POSTERS**
	Session 1 – *Auditorium, L1* *Chairs: Ms Cindy Zaitsoff & Dr Rebekah McWhirter* Screening strategies for recruitment and result reporting to maximise utility of whole-of-life genomics **– *Prof Leslie Burnett*** Improving communication of genetic results in families with hypertrophic cardiomyopathy: a randomised controlled trial **– *Miss Charlotte Burns*** "The benefit that comes from my existence": Patients' experiences of consenting to a cancer rapid autopsy program **– *Dr Laura Forrest*** Pregnancy outcomes following the detection of early fetal edema on pre-NIPT ultrasound **– *Dr Melody Menezes*** The expanding LARS2 phenotypic spectrum: HLASA, Perrault syndrome with leukodystrophy, and mitochondrial myopathy **– *Dr Lisa Riley***

MONDAY 5 AUGUST 2019	
	Session 2 – *Frank Taplin Room, L1* *Chairs: Prof Martin Delatycki & Dr Bryony Ryder* What ExACtly do we know about Functional Constraint on Genes Associated with End-Stage Kidney Failure? **– *Ms Hope Tanudisastro*** Western Australian health professionals' attitudes to and knowledge about expanded preconception carrier screening **– *Mr Royston Ong*** Modelling the mitochondrial disease Sengers syndrome using human embryonic stem cells **– *Mr Yau Chung Low*** The practice of engaging Aboriginal and Torres Strait Islander communities in genome research **– *Ms Azure Hermes*** Breast screening in young women with NF1: Psychological impact and development of an educational resource **– *Ms Ashley Crook***
	Session 3 – *Lion Harbour View, L2* *Chairs: Prof Clara Gaff & Prof John Christodoulou* Biallelic variants in EFEMP1 in a man with a pronounced connective tissue phenotype **– *Dr Emma Wade*** ROSAH syndrome: an autosomal dominant ocular and multisystem disorder with causative variant, ALPK1 p.Thr237Met **– *Dr Amin Sabri*** Telomere length in skeletal muscle and leukocytes, and aerobic fitness **– *Dr Danielle Hiam*** A Novel Method for Gene and Region Prioritization Based on Human Phenotype Ontology (HPO) Terms **– *Mr Brian Lee*** Early-career Doctor recommendations for competencies in genomics **– *A/Prof Diane Kenwright***
1415-1600	**Plenary & Themed Session 4: Community Engagement** – *Auditorium, L1* *Chairs: Dr Danya Vears & Prof Stephen Robertson* Plenary 11: Enhancing responsibility in research and response-ability in practice **– *A/Prof Maui Hudson*** Plenary 12: Indigenous people and genomics in Australia **– *Prof Emma Kowal*** Plenary 13: Engaging with Indigenous Communities for genetic research: Benefits, Pitfalls and Lessons **– *Dr Rebekah McWhirter*** Plenary 14: Te Aho Matatū – Culturally responsive genetic research **– *Dr Karyn Paringatai*** Panel Discussion
1600-1630	**AFTERNOON TEA & EXHIBITION** – *Renouf Foyer & Lower Promenade, L1*

MONDAY 5 AUGUST 2019	
CONCURRENT SESSIONS - SUBMITTED ORALS (TOP 15 ORALS)	
1630–1745	**Orals – Session 1** – *Auditorium, L1* *Chairs: A/Prof Veronica Wiley & Ms Alice Christian* Results of the North Carolina Newborn Exome Sequencing for universal screening (NC NEXUS) study **– *Prof Cynthia Powell (invited speaker)*** The changing nature of genomic medicine: Australian medical specialists' current practice, preparedness and preferences **– *Dr Amy Nisselle, Dr Belinda McClaren*** Hearing their voices: The economic and psychosocial impacts on families affected by intellectual disability **– *Prof Deborah Schofield*** Preparing for Mackenzie's Mission and Expanded Carrier Screening at Victorian Clinical Genetic Services (VCGS) **– *Dr Justine Marum*** Parent decision making about clinical trials for Fragile X Syndrome **– *Dr Erin Turbitt***
	Orals – Session 2 – *Frank Taplin Room, L1* *Chairs: Ms Kimberly Gamet & Dr Sarah Heron* Homozygous inheritance of AAGGG RE in RFC1 causes CANVAS **– *Dr Haloom Rafehi*** Genetic and cellular characterisation of cortical dysplasia using patient-derived brain tissue **– *Dr Paul Lockhart*** Pathogenic variants in GPC4 cause Keipert syndrome **– *Dr David Amor*** Human sex reversal is caused by duplication or deletion of core enhancers upstream of SOX9 **– *Prof Andrew Sinclair*** Multi-gene analysis effectively provides a high diagnostic yield and resolves differential diagnoses in neuromuscular disorders **– *Dr Swaroop Aradhya***
	Orals – Session 3 – *Lion Harbour View, L2* *Chairs: Dr Michael Gabbett & Ms Louise Carey* Evaluating cost-effectiveness of exome sequencing in a prospective versus historical cohort of complex paediatric patients **– *Dr Alison Yeung*** Implementing clinically validated automated genomic variant prioritisation with diagnostic performance that equals human experts **– *Prof Leslie Burnett*** A national approach to rapid genomic diagnosis in acute paediatrics **– *A/Prof Zornitza Stark*** Implementation of exome sequencing in a clinical diagnostic laboratory service **– *Ms Justine Elliott*** Exome sequencing enhances the diagnostic rate of perinatal autopsy: A prospective multicentre clinical utility trial with implications for prenatal diagnosis **– *Dr George Mcgillivray***
1900–2330	**CONFERENCE DINNER** *Museum of New Zealand, Te Papa Tongarewa*

TUESDAY 6 AUGUST 2019		
0800-1300	**REGISTRATION DESK** – *Fletcher Challenge Foyer, Grd Fl*	
0830-1000	**PLENARY & THEMED SESSION 5: THERAPIES** - *Auditorium, L1* *Chairs: Dr Emma Glamuzina & A/Prof Kathy Tucker* Plenary 15: Assessing the effects of new small molecule-based treatments **– *Prof Peter Clayton*** Plenary 16: Developments in the use of disease-modifying treatments to prevent or reverse the neurodegenerative process in HD **– *Dr Edward Wild*** Plenary 17: Drug trials in skeletal dysplasia **– *Prof Ravi Savarirayan*** Plenary 18: Therapeutic advances in the management of mitochondrial disorders **– *Prof John Christodoulou***	
1000-1100	**MORNING TEA & EXHIBITION** Exhibition & Posters close at the end of morning tea. *Renouf Foyer, Lower Promenade, Fletcher Challenge Foyer*	**1015-1100** *Lion Harbour View, L2* **Sophia Genetics Industry Session** **SOPHiA Whole Exome Solution: reducing turnaround time, increasing diagnostic yield** ***Ms Giorgia Riboldi-Tunnicliffe*** SOPHiA™ SOPHIAGENETICS.COM
1100-1145	**HGSA ORATION** - *Auditorium, L1* *Chair: Dr Michael Buckley* Systems and Change ***Ms Mary-Anne Young***	
1145-1245	**HGSA AGM & CERTIFICATE PRESENTATIONS** – *Auditorium, L1*	
1245-1300	**2020 ASM PRESENTATION** – *Auditorium, L1*	
1300-1315	**CLOSING, ASGC 25TH ANNIVERSARY PRESENTATION & AWARD PRESENTATIONS** – *Auditorium, L1*	
1315-1400	**CLOSING LUNCH** – *Fletcher Challenge Foyer*	