15th GeneMappers Conference 2021 Program

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Wednesday 23rd of June

Session (Chair)	Speaker	Title	Time UTC 10
Arrival/Preparation. **IF YOU ARE ATTENDING A LOCAL HUB** you must arrive with enough time to complete COVID-safe registration and checking in.			12:20-13:00
Conference Welcome	Prof. Eske Derks, Assoc. Prof. Matthew Law & Dr. Nicole Warrington	Welcome and opening comments. Acknowledgement of country will be delivered by Gregory Pratt, manager of Aboriginal and Torres Strait Islander Health Research at QIMR	13:00-13:10
Invited speaker #1 (Assoc. Prof. Matthew Law**)**	**Prof. Melanie Bahlo**	Renaissance of repeat expansions: more than just a monogenic spectacle	13:10-13:40
Session 1: Functional genomics (Assoc. Prof. Tracy O'Mara)	**Speaker**	**Title**	**13:40-15:00**
	Yang Wu	Joint analysis of multi-omics data reveals molecular mechanisms at GWAS loci	13:40-13:55
	Brittany Mitchell	Genome-wide association meta-analysis of acne reveals novel risk loci and molecular overlap with breast cancer	13:55-14:10
	Sarahi Rivera	Integrating GWAS and 3D chromatin interactome data to identify multi-cancer risk genes in hormone-related cancers	14:10-14:25
	Ting Qi	Genetic control of RNA splicing and its distinctive role in complex trait variation	14:25-14:40
	Alan Rubin	Accessing functional assay data for your favorite genes via MaveDB	14:40-14:55
Afternoon tea 1			15:00-15:30
Session (Chair)	**Speaker**	**Title**	**Time UTC 10**
Invited speaker #2 (Prof. Murray Cairns)	**Dr. Sonia Shah**	Using genomics to investigate the effects of statins on depression	15:30-16:00
Invited speaker #3 (Prof. Eske Derks)	**Prof. Danielle Posthuma**	From GWAS to function	16:00-16:40
Session 2: GWAS and complex disease genetics I (Dr. Zachary Gerring)	**Speaker**	**Title**	**16:40-17:40**
	Swapnil Tichkule	Genomic landscape of diversification, selective sweeps and demographic history in an anthroponotic parasite	16:40-16:55
	Sally Mortlock	Unravelling disease heterogeneity using genetics: The relationship between endometriosis and ovarian cancer subtypes.	16:55-17:10
	Danielle Adams	Investigating exposure mediated genetic scoring for coronary artery disease	17:10-17:25
	Matthew Wakefield	Testing all possible mutations in PARP1 to prospectively identify resistance mutations and drug substitutions that will overcome resistance	17:25-17:40
Close of day 1	Prof. Eske Derks , Assoc. Prof. Matthew Law & Dr. Nicole Warrington	Closing comments, outstanding business	17:40-18:00

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Thursday 24th of June

Session (Chair)	Speaker	Title	Time UTC 10
Arrival 2			07:30-08:00
Invited Speaker #4 (Prof. Sarah Medland)	**Prof. Benjamin Neale**	Progress in Understanding the Genetic Basis of Mental Illnesses	08:00-08:40
Session 3: Analysis of low-frequency variants (Dr. Nicholas Blackburn)	**Speaker**	**Title**	**08:40-10:00**
	Huanwei Wang	Quantification of the contribution of ultra-rare coding variants to trait heritability	08:40-08:55
	Erandee Robertson	A hidden Markov model to identify inherited disease-causing mutations using shared genetic markers	08:55-09:10
	Paul Dunn	Mitochondrial mutations identify links between cerebral small vessel disease and Alzheimer's disease	09:10-09:25
	Antony Kaspi	Identifying genetic causes of childhood apraxia of speech using whole genome high throughput sequencing	09:25-09:40
	Kim Ngan Tran	Multi-phenotype genome-wide association studies of the Norfolk Island isolate implicate pleiotropic loci involved in chronic kidney disease	09:40-09:55
Morning Tea 1			10:00-10:30
Invited Speaker #5 (Dr. Carol Dobson-Stone)	**Assoc. Prof. Mark Cowley**	Integrating Whole Genome and RNA sequencing for national-scale precision medicine for children with cancer	10:30-11:00
Session 4: GWAS and complex disease genetics II (Prof. David Evans)	**Speaker**	**Title**	11:00-12:00
	Md Rafiqul Islam	Exploring the genetic relationship between migraine and glucose-related traits	11:00-11:15
	Jackson Thorp	Symptom-level genetic modelling identifies novel risk loci and unravels the shared genetic architecture of anxiety and depression	11:15-11:30
	Evgeniia Golovina	Autism spectrum disorder: understanding the impacts of SNPs on biological pathways in the human fetal and adult cortex.	11:30-11:45
	Jacqueline Kiewa	Distinct profile of metabolite polygenic risk scores may distinguish perinatal depression from major depressive disorder	11:45-12:00
Lunch			12:00-13:00
Invited Speaker #6 (Dr. Julia Steinberg)	**Prof. Anne Cust**	Results from the Melanoma Genomics Managing Your Risk Study – a PRS-based RCT assessing impact on behaviours and psychological outcomes	13:00-13:30

Thursday 24th of June cont.

	Speaker	Title	13:30-14:00
Poster Lightning Session I (Assoc. Prof. Matthew Law)	Anna Freydenzon	Comparative RNA-seq reveals upregulated tissue generation in skeletal muscle of amyotrophic lateral sclerosis cases to controls	13:30-13:34
	Geoffrey English	Polygenic risk for insomnia significantly predicts hyperactivity/impulsivity but not inattention	13:34-13:38
	Enda Byrne	The Australian Genetics of Depression Study: new risk loci and dissecting heterogeneity between subtypes	13:38-13:42
	Richard Campbell	Identification of inter-chromosomal SNP varian signatures with extreme effects in human disease from GWAS and validation in the UK-Biobank	13:42-13:46
	Victoria Jackson	Initial findings from the International Genetics of Stuttering Study	13:46-13:50
	Jue Sheng Ong	Evaluating the complex relationship between balding and skin cancer in men through multivariable Mendelian randomization analyses	13:50-13:54
Poster Lightning Session II (Prof. Eske Derks)	**Speaker**	**Title**	**14:00-14:30**
	Liliana Ciobanu	Mapping the genetic architectures of cognitive functioning	14:00-14:04
	Jose Morosoli	Literacy challenges for science communication and complex trait genetics: Insights from text mining and survey studies	14:04-14:08
	Xiaodong Mo	Prediction of heart failure using genetic information and traditional risk factors in UK biobank	14:08-14:12
	Michael Geaghan	Investigating the enrichment of microRNA binding site variants in psychiatric disorders	14:12-14:16
	Karen Mather	Discovering putative protein biomarkers for successful ageing using polygenic risk scores and the exceptional longevity model	14:16-14:20
	Ang Li	A novel set-based association test improves power of identifying loci harbouring variants with masking effects	14:20-14:24

Thursday 24th of June cont.

Poster Lightning Session III (Dr. Nicole Warrington)	**Speaker**	**Title**	**14:30-15:00**
	Alesha Hatton	Characterising the trans-ancestral genetic control of DNA methylation	14:30-14:34
	Guiyan Ni	Fluctuating asymmetry of bilateral traits is associated with disease and reproductive fitness in humans	14:34-14:38
	Maria Pia Campagna	Whole-blood methylation as a prognostic biomarker in multiple sclerosis	14:38-14:42
	Patrick Wang	Multi-trait Bayesian genome-wide association study identifies novel endometrial cancer risk loci at 7q22.1, 8q24.3, and 16q22.2	14:42-14:46
	William Reay	Genetic support for drug repurposing through integration of causal inference, transcriptomic imputation, and polygenic scoring	14:46-14:50
	Longfei Wang	Mitochondrial DNA copy number inferred from next generation sequencing data is higher in individuals with autism spectrum	14:50-14:54
Afternoon tea 2			15:00-15:30
Invited Speaker #7 (Dr. Saskia Freytag)	**Prof. Ryan Lister**	Unveiling hidden cellular states and their regulators through single cell genomics	15:30-16:00
New Technology Talk (Assoc. Prof. Matthew Law)	**Illumina Platinum Sponsor**	**New Technology Talk**	16:00-16:15
Session 5: Methods development and application (Dr. Victoria Jackson)	**Speaker**	**Title**	**16:15-17:15**
	Anubhav Kaphle	Developing a better model for SNP-heritability analysis and genomic prediction	16:15-16:30
	Yuanxiang Zhang	Detection of assortative mating in the absence of spousal information	16:30-16:45
	Valentin Hivert	Estimation of non-additive genetic variance in human complex traits from a large sample of unrelated individuals	16:45-17:00
	Daniel Hwang	Using adopted individuals to partition materna genetic effects into pre and post-natal effects on offspring phenotype	17:00-17:15
Close of day 2	Prof. Eske Derks , Assoc. Prof. Matthew Law & Dr. Nicole Warrington -	Closing comments, outstanding business	17:15-17:20
Recast 1		Recast of Invited Speaker #4 and Session 3	17:20-19:20

Friday 25th of June

Session (Chair)	Speaker	Title	Time UTC 10
Arrival 3			**08:30-09:00**
Session 6: GWAS and complex disease genetics III (Assoc. Prof. Sarah Cohen-Woods)	**Speaker**	**Title**	**09:00-10:00**
	Weixiong He	Multi-trait genome-wide association study identifies novel loci associated with corneal traits and keratoconus	09:00-09:15
	Puya Gharahkhani	Genetic factors explain varying glaucoma prevalence and pressure subtype across ancestries	09:15-09:30
	Fei-Fei Cheng	Genome-wide Association Study and Polygenic Risk Prediction for Refractive Error	09:30-09:45
	Dylan Kiltschewskij	Dissecting Genetic Correlation and Causation Amongst Biochemical Traits and Cortical Properties	09:45-10:00
Morning Tea 2			**10:00-10:30**
Invited Speaker #8 (Prof. Martin Kennedy)	**Prof. Nigel Laing**	Diagnosing genetic disease before it happens: carrier screening and Mackenzie's Mission	10:30-11:10
Invited Speaker #9 (Dr. Loic Yengo)	**Assis. Prof. Po-Ru Loh**	Protein-altering variants with large phenotypic effects: rare SNPs and common VNTRs	11:10-11:50
Conference close	Prof. Eske Derks , Assoc. Prof. Matthew Law & Dr. Nicole Warrington	**Prize awards.** Planning for next GeneMappers. Closing remarks.	11:50-12:30
Recast 2		**Recast of Session 6**	**12:30-13:30**

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Online posters

Abstract	Presenter	Title
7	Jodie Painter	Miscarriage and the risk of psychological distress
8	Natalie Twine	Revolutionising polygenic risk models using machine learning
10	Shannon D'Urso	Mendelian Randomization Analysis of Fertility Phenotypes and Endometrial Cancer Risk in the UK-Biobank
11	Zachary Gerring	The local genetic overlap of psychiatric and substance use phenotypes
20	Lyndal Henden	Identity by descent analysis links global MND and FTD cases for disease gene discovery and supports neurodegenerative disease spectrum
22	Ammarah Ghaffar	Genome-wide Imputation Differential Expression Enrichment
28	Thao Van Cao	A 10-year longitudinal study of epigenomics and body fat traits in Norfolk Island population
29	Toyin Abdulsalam	Identification of blood gene expression quantitative trait loci (eQTLs) in older adults
30	Mischa Lundberg	Genetic, lifestyle and environmental risk factors for chronic pain revealed through GWAS
31	Nathan Ingold	Leveraging power from multiple genetically correlated traits to identify genetic loci associated with perceived aging
34	Mary Revelas	High polygenic risk score for exceptional longevity is associated with a healthy metabolic profile
41	Mathias Seviiri	A multi-phenotype analysis reveals 21 novel susceptibility loci for basal cell carcinoma and 18 for squamous cell carcinoma.
44	Ebrahim Mahmoudi	Genome-wide investigation of VNTRs in schizophrenia
46	Beilei Bian	The role of critical immune genes in brain disorders: insights from neuroimaging immunogenetics
47	Sri Chandana Kanchibhotla	Moderate Heritability of Peripheral Blood Gene Expression in Older Adults
48	Julia Steinberg	Genomic testing to identify colorectal cancer patients with Lynch syndrome: current practice and gaps in seven Australian hospitals
50	Kathryn Kemper	Genetic control of intra-individual variability
51	Joana Revez	Cross-ancestry analyses of 25 hydroxyvitamin D levels in the UK Biobank
55	Sulagna Banerjee	A Xenopus laevis epilepsy model to test alternative therapy for Intractable Epilepsy
56	Bianca Grosz	Investigating the Pathogenic Mechanism of the Non-coding c.-103C>T GJB1 5' UTR Mutation in CMTX1

Abstract	Presenter	Title
59	Caitlin King	Comparing clinical genetics research involving Aboriginal, Torres Strait Islander and Maori populations from 2000 to 2020.
60	Geng Wang	Investigating a potential causal relationship between maternal blood pressure during pregnancy and future offspring cardiometabolic health
67	Omar Ibrahim	Machine Learning accurately predicts severity of multiple sclerosis where GWAS fails
68	Kelsie Raspin	A rare germline variant in the DNA damage repair gene RAD51C is associated with familial prostate cancer risk
69	Georgea Foley	A rare genetic variant in OR5H14 is associated with familial prostate cancer risk
70	Adam Kowalczyk	Explicit signals of joint association significantly improve genomic prediction of height and total sugar in a population of sorghum plants
72	Sionne Lucas	Identifying putative disease-causing variants using whole genome sequencing in families with multiple cases of idiopathic pulmonary fibrosis