

# Symposium Program



<b>Wednesday</b>				
<b>March 25th</b>				
	<i>Time</i>	<i>Speaker</i>	<i>Institution</i>	<i>Title</i>
	<b>10:00-10:15</b>	<b>Welcome to Country</b>		
<b>Session 1</b>	<b>10:15-11:00</b>	<b>Fumitaka Inoue</b>	<b>Kyoto University</b>	<b>Deciphering human functional genome using Massively Parallel Reporter Assays</b>
Chair: Irene Gallego Romero				
	11:00-11:15	Chanodya Pasansi Ranwala	University of Adelaide	Unraveling Regulatory Complexity: Functional Characterisation of Enhancer Variants associated with Common Epilepsy
	11:15-11:30	Asfar Lathif Salaudeen	The University of British Columbia	Expanding the gene editing toolkit to decipher endogenous causal variants
	<b>11:30-11:45</b>	<b>Coffee break</b>		
<b>Session 2</b>	11:45-12:00	Daniel V Brown	Walter and Eliza Hall institute of Medical Research	Active learning of the sequence-function landscape in a far-red fluorescent protein
Chair: Lachlan Jolly				
	12:00-12:15	Maddy Comerford	University of Melbourne	Mapping the gene regulatory landscape of archaic hominin introgression in modern Papuans
	12:15-12:30	Justine Shih	Broad Institute of MIT and Harvard	Mapping Genetic Variation to Cellular Phenotypes with Targeted Optical Endogenous Sequencing
Lara Muffley	12:30-12:45	Bernd Willems	Twist Bioscience	Writing the Future of Mutational Scanning
	<b>12:45-13:45</b>	<b>Lunch + posters on your own</b>		
<b>Session 3</b>	<b>13:45-14:15</b>	<b>Bryony Thompson</b>	<b>Royal Melbourne Hospital</b>	<b>Lessons Learned from Applying Functional Data in Real-World Clinical Variant Classification</b>
Chair: Mandy Spurdle				
	14:15-14:30	Melissa Gilbert	The Children's Hospital of Philadelphia & University of Pennsylvania	The clinical utility of Multiplexed Assays of Variant Effects (MAVEs) in a pediatric cohort
	14:30-14:45	Richard James	Seattle Children's Research Institute	Functional Assessment of Genetic Variants of CARD11 and BCL10 with Saturation Genome Editing
	14:45-15:00	Ben Capodanno	Brotman Baty Institute at the University of Washington	MaveMD: A functional data resource for genomic medicine
	15:00-15:15	Emmylou C. Nicolas-Martinez	Adelaide University	RNA variants resolved in non-expressed genes by transactivation
	<b>15:15-15:45</b>	<b>Coffee Break</b>		
<b>Session 4</b>	<b>15:45-16:15</b>	<b>Alex Wagner</b>	<b>Nationwide Children's Hospital</b>	<b>From Functional Assays to Computable Knowledge: Making MAVE Data Usable in Variant Interpretation</b>
Chair: John Christodoulos				
	16:15-16:30	Steven E. Brenner	University of California, Berkeley	Findings from the Critical Assessment of Genome Interpretation (CAGI), seventh edition: a community experiment to evaluate phenotype prediction
	16:30-16:45	Ashley P.L. Marsh	Ambry Genetics	Clinical translation of a MUTYH MAVE into a diagnostic laboratory workflow enables large-scale VUS resolution
	16:45-17:00	Frederick P. Roth	University of Pittsburgh	Landscapes of human AAT missense-variant effects reveal pathogenic variation and genetic suppressors

<b>Thursday</b>				
<b>March 26th</b>				
	<i>Time</i>	<i>Speaker</i>	<i>Institution</i>	<i>Title</i>
<b>Session 5:</b>	<b>9:00-9:45</b>	<b>Matthew Hurles</b>	<b>Wellcome Sanger Institute</b>	<b>Keynote Speaker - Scaling MAVES: are we at the end of the beginning?</b>
<b>Chair: Rehan Villani</b>	<b>9:45-10:15</b>	<b>Chai-Ann Ng</b>	<b>Victor Chang Cardiac Research Institute</b>	<b>Integrating MAVE and patch clamp data to improve classification and risk prediction in KCNH2-LQTS</b>
	10:15-10:30	Andrew Glazer	Vanderbilt University Medical Center	Three Multimodal Assays of SCN5A Variant Function Inform Arrhythmia Risk Prediction
	10:30-10:45	Sujatha Jagannathan	University of Colorado Anschutz Medical Campus	Single-codon resolution mapping of nonsense-mediated mRNA decay via genomic stop codon scanning of LMNA
	10:45-11:00	Lea Starita	University of Washington	260,000 variant effect measurements and novel calibration methods drive resolution of VUS
	<b>11:00-11:30</b>	<b>Coffee Break</b>		
<b>Session 6:</b>	<b>11:30-12:00</b>	<b>Nilah Ioannidis</b>	<b>University of California, Santa Cruz</b>	<b>Modeling the impact of personal genome variation on molecular phenotypes</b>
Chair: Sandra Cooper	12:00-12:15	Omar Tariq	University of British Columbia	tfGPRA: A High Throughput Platform for Transcription Factor Characterization
	12:15-12:30	Vanessa Burns	Wellcome Sanger Institute	Saturation Genome Editing to Clarify Variant Effect within 5'UTRs of Neurodevelopmental Disorder Genes
	12:30-12:45	Lorenzo Vaccaro	Telethon Institute of Genetics and Medicine	Genotype-phenotype single-cell transcriptomics for massive parallel assessment of genetic variants
Lara Muffley	12:45-13:00	Daniel Zimmerman	Ambry Genetics	MAVE Progress Report, How Ambry Genetics Validates and Deploys High-Throughput Functional Assays
	<b>13:00-14:00</b>	<b>Lunch + posters on your own</b>		
<b>Session 7:</b>	<b>14:00-14:30</b>	<b>Jian-Rong Yang</b>	<b>Sun Yat-sen University, Guangzhou</b>	<b>Phenotypic Mutations in Collision: Negative Epistasis Between Transcription Errors and Translation Errors Revealed by DMS</b>
Chair: Matthew Wakefield	14:30-14:45	Srivatsan Raman	University of Wisconsin-Madison	From Variant Maps to Functional Design: Using Deep Mutational Scanning and Machine Learning to Engineer Programmable Bacteriophages
	14:45-15:00	Guillaume Diss	Friedrich Miescher Institute	The genetic architecture of the human bZIP interaction network
	15:00-15:15	Jacob Purcell	Monash University	High-resolution functional assessment of SMAD4 variants
	15:15-15:30	Rosa De Santis	Telethon Institute of Genetics and Medicine	From bulk to single cell: benchmarking analytical tools for Deep Mutational Scanning experiments
	15:30-16:00	<b>Coffee Break</b>		
<b>Session 8:</b>	<b>16:00-16:30</b>	<b>Amelie Stein (v)</b>	<b>University of Copenhagen</b>	<b>Variant consequences in context</b>
Chair: Sefi Rosenbluh	16:30-16:45	Xinyu Wu	Walter and Eliza Hall institute of Medical Research	Elucidate the active conformation of cytokine receptors using deep mutational scanning
	16:45-17:00	Polina V Polunina	University of Freiburg	Modeling Viral Evolution as Sequence Transitions: A Transformer Approach Using Time-Resolved SARS-CoV-2 Spike Data
Fritz Roth	17:00-17:30	Selected poster short talks		
	<b>17:30-19:30</b>	<b>Catered poster session</b>		
	<b>19:45-late</b>	<b>Social function at the Gertrude Hotel (148 Gertrude St, five minute walk)</b>		

<b>Friday</b>				
<b>March 27th</b>				
	<i>Time</i>	<i>Speaker</i>	<i>Institution</i>	<i>Title</i>
<b>Session 9:</b>	<b>9:00-9:45</b>	<b>Maitreya Dunham</b>	<b>University of Washington</b>	<b>Keynote Speaker: From ARS1 to ZWFI: some adventures in deep mutational scanning</b>
Chair: Alan Rubin	<b>9:45-10:15</b>	<b>Mark Dawson</b>	<b>Peter MacCallum Cancer Centre</b>	<b>Characterising the functional landscape of the human PRC2 complex by base editing at single cell resolution</b>
	10:15-10:30	Maximilian Stamnitz (v)	Centre for Genomic Regulation	The genetic architecture of allosteric plant hormone receptors
	10:30-10:45	Jun J Yang	St. Jude Children's Research Hospital	MAVE-informed Protein Language Models for Predicting Pharmacogenetic Variant Function at Scale
	10:45-11:00	Douglas M. Fowler	University of Washington	Biochemical profiling of ~315,000 MAP kinase pathway protein variants in human cells with LABEL-seq
	<b>11:00-11:30</b>	<b>Coffee Break</b>		
<b>Session 10:</b>	<b>11:30-12:00</b>	<b>Chris Hahn</b>	<b>SA Pathology and University of South Australia</b>	<b>Deep mutational scanning to measure the transactivational effect of GATA2 and ERG variants to benefit clinical interpretation</b>
Chair: Ebony Matotek	12:00-12:15	Alistair Dunham	Wellcome Sanger Institute	Standardisation and joint analysis of 2,041 MAVEs
	12:15-12:30	Yu-Jen (Jennifer) Lin	University of California, Berkeley	Critical Assessment of Genome Interpretation (CAGI) 7 ARSA missense stability prediction challenge identifies computational advances over state-of-the-art variant impact predictors
	12:30-12:45	Linda Wijaya	The Kids Research Institute Australia & University of Western Australia	Unravelling the RASopathy Syndromes using iPSC-derived neural disease modelling
Lara	12:45-13:00	TBC	Illumina Inc	TBC
	<b>13:00-14:00</b>	<b>Lunch (no posters)</b>		
<b>Session 11:</b>				
Chair: Convenors	<b>14:00-14:30</b>	<b>Melissa Call</b>	<b>Walter and Eliza Hall Institute of Medical Research</b>	<b>Mutational profiling of SARS-CoV-2 PLpro exposes inhibitor escape routes</b>
Convenors	14:30-14:45	Awards and closing		
		Coffee Break		
	15:00-17:00	<b>Workshop</b>		Clinical Application Workshop