DRAFT Symposium Program

MSS 2026
Mutational Scanning Symposium

Wednesday			
March 25th			
Time	*Speaker*	*Institution*	*Title*
10:00-10:15	**Welcome to Country**		
10:15-11:00	**Fumitaka Inoue**	**Kyoto University**	***Deciphering human functional genome using Massively Parallel Reporter Assays***
11:00-11:15	Chanodya Pasansi Ranwala	Adelaide, Australia	Unraveling Regulatory Complexity: Functional Chracterisation 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
11:30-11:45	**Coffee break**		
11:45-12:00	Daniel V Brown	Walter and Eliza Hall Institute	Active learning of the sequence-function landscape in a far-red fluorescent protein
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
12:30-12:45	Twist Biosciences	Bernd Willems	TBC
12:45-13:45	**Lunch + posters on your own**		
13:45-14:15	**Bryony Thompson**	**Royal Melbourne Hospital**	**Lessons Learned from Applying Functional Data in Real-World Clinical Variant Classification**
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	Jeremy Stone	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, Adelaide, Australia	RNA variants resolved in non-expressed genes by transactivation
15:15-15:45	**Coffee Break**		
15:45-16:15	**Alex Wagner**	**Nationwide Children's Hospital**	**TBC**
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 Corporation, Aliso Viejo, CA	Clinical translation of a MUTYH MAVE into a diagnostic laboratory workflow enables large-scale VUS resolution
16:45-17:00	Frederick P. Roth	Pittsburgh, PA, USA	Landscapes of human AAT missense-variant effects reveal pathogenic variation and genetic suppressors

Thursday			
March 26th			
Time	*Speaker*	*Institution*	*Title*
9:00-9:45	**Matthew Hurles**	**Wellcome Sanger Institute**	***Keynote Speaker - Title TBC***
9:45-10:15	**Chai-Ann Ng**	**Victor Chang Cardiac Research Institute**	**Integrating MAVE and patch clamp data to improve classification and risk prediction in KCNH2-LQTS**
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, Aurora, CO, USA	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
11:00-11:30	**Coffee Break**		
11:30-12:00	**Nilah Ioannidis**	**University of California, Santa Cruz**	**Modeling the impact of personal genome variation on molecular phenotypes**
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, Department of Translational Medicine	Genotype-phenotype single-cell transcriptomics for massive parallel assessment of genetic variants
12:45-13:00	Daniel Zimmerman	Ambry Genetics	MAVE Progress Report, How Ambry Genetics Validates and Deploys High-Throughput Functional Assays
13:00-14:00	**Lunch + posters on your own**		
14:00-14:30	**Jian-Rong Yang**	**Sun Yat-sen University, Guangzhou**	**Phenotypic Mutations in Collision: Negative Epistasis Between Transcription Errors and Translation Errors Revealed by DMS**
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	Maximilian Stammnitz	Centre for Genomic Regulation	The genetic architecture of allosteric plant hormone receptors
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	**Coffee Break**		
16:00-16:30	**Chris Hahn**	**SA Pathology and University of South Australia**	**Deep mutational scanning to measure the transactivational effect of GATA2 variants to benefit clinical interpretation**
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
17:00-17:30	Selected poster short talks		
17:30-19:30	**Catered poster reception**		

Friday			
March 27th			
Time	*Speaker*	*Institution*	*Title*
9:00-9:45	**Maitreya Dunham**	**University of Washington**	***Keynote Speaker: From ARS1 to ZWF1: some adventures in deep mutational scanning***
9:45-10:15	**Mark Dawson**	**Peter MacCallum Cancer Centre**	**TBC**
10:15-10:30	Jun J Yang	St. Jude Children's Research Hospital	MAVE-informed Protein Language Models for Predicting Pharmacogenetic Variant Function at Scale
10:30-10:45	Jacob Purcell	Monash University	High-resolution functional assessment of SMAD4 variants
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
11:00-11:30	**Coffee Break**		
11:30-12:00	**Amelie Stein**	**University of Copenhagen**	**TBC**
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	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	Brendan Larsen	Fred Hutch Cancer Center	Functional and antigenic landscape of the Nipah virus entry proteins
12:45-13:00	Linda Wijaya	The Kids Research Institute Australia, University of Western Australia	Unravelling the RASopathy Syndromes using iPSC-derived neural disease modelling
13:00-14:00	**Lunch (no posters)**		
14:00-14:30	**Melissa Call**	**Walter & Eliza Hall Institute**	**TBC**
14:30-14:45	Awards and closing		
	Coffee Break		
15:00-17:00	**Workshop**		Clinical Application Workshop