

Human Genome Meeting 2020







5-8 April 2020
Virtual Meeting

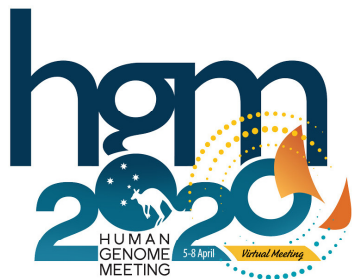
All times listed for Perth, Australia (+8h GMT). Please check your local time zone for individual requirements.

HGM Early Career Researcher Symposium SUNDAY 5 APRIL 2020

Our thanks to Symposium Sponsor



| 08:30 – 10:05 | SESSION 1 <i>Chair: Miss Sarah Beecroft</i> | Channel Sponsor  |
|---------------|--|--|
| 08:30- 08:35 | WELCOME Prof Alistair Forrest <i>Convenor HGM2020</i> | |
| | ECR COMMITTEE WELCOME | |
| 08:35 – 09:05 | MENTORING SPEAKER Prof Daniel MacArthur <i>Broad Institute of MIT and Harvard, United States</i> | |
| 09:05 - 09:15 | CORRELATIVE AND CAUSAL RELATIONSHIPS BETWEEN COMPLEX TRAITS AND BRAIN VOLUMES ARE MASKED BY EDUCATION AND SOCIOECONOMIC STATUS Dr Frank Wendt <i>Yale School of Medicine, United States</i> | |
| 09:15 – 09:25 | DEVELOPMENT OF COMBINATORIAL EPIGENOME EDITING TECHNOLOGIES THROUGH A MASSIVELY PARALLELIZED SINGLE CELL RNA-SEQ SCREEN Dr Christian Pflueger <i>The University of Western Australia, Australia</i> |  |
| 09:25 – 09:35 | GENETIC HETEROGENEITY OF POLYMICROGYRIA: STUDY OF 124 PATIENTS USING DEEP SEQUENCING Dr Chloe Stutterd <i>MCRI, Australia</i> |  |
| 09:35 – 09:45 | RESCUE OF FIBRILLIN-1 MICROFIBRIL FORMATION IN FIBROBLASTS FROM INDIVIDUALS WITH MARFAN SYNDROME Ms Jessica Cale <i>Murdoch University, Australia</i> |  |
| 09:45 – 09:55 | CHARACTERIZING THE SPATIAL HETEROGENEITY OF TUMOUR TISSUES BY IDENTIFYING CANCER AND IMMUNE CELL-TYPES, CELL-SUBTYPE DISTRIBUTION AND TRANSCRIPTIONAL REGULATION Mr Duy Pham <i>Institute for Molecular Bioscience, University of Queensland, Australia</i> |  |
| 09:55 – 10:05 | INTEGRATING RNA-SEQ WITH ENVIRONMENT EXPOSURE FOR PRIORITIZATION OF DIAGNOSIS BIOMARKERS WITH SCHIZOPHRENIA Dr Jiahui Ma <i>Peking University, China</i> |  |
| 10:05 – 10:40 | BREAK | |

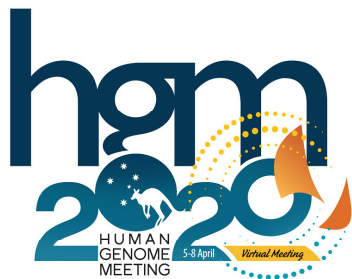


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| 10:40 – 11:50 | SESSION 2 <i>Chair: Miss Shelley Watters</i> | Channel Sponsor  |
|---------------|---|--|
| 10:40 – 10:50 | ANTISENSE OLIGONUCLEOTIDE-MEDIATED EXON SKIPPING TO TREAT SPINOCEREBELLAR ATAXIA TYPE 3 Mr Craig McIntosh <i>CMMIT, Murdoch University, Australia</i> |  |
| 10:50 – 11:00 | DISTINCT TRANSCRIPTOMIC PROFILES FOR ACTIVATED AND RESTING T FOLLICULAR HELPER CELLS FOLLOWING A SUCCESSFUL RESPONSE TO THE INFLUENZA VACCINE Mx Jennifer Currenti <i>University of Western Australia, Australia</i> |  |
| 11:00 – 11:10 | GENETIC LEGACY OF THE LAST HUNTER-GATHERER GROUP IN BORNEO Dr Pradiptajati Kusuma <i>Eijkman Institute for Molecular Biology, Singapore</i> | |
| 11:10 – 11:20 | GENERATION AND TRANSCRIPT PROFILING OF IPSC, RETINAL ORGANOID AND RPE IN RCBTB1-RELATED RETINOPATHY Ms Zhiqin Huang <i>Lions Eye Institute, Australia</i> |  |
| 11:20 – 11:30 | NATMI: A NETWORK ANALYSIS TOOLKIT FOR MULTICELLULAR INTERACTIONS IN SINGLE-CELL TRANSCRIPTOME DATA Mr Rui Hou <i>Harry Perkins Institute of Medical Research, Australia</i> |  |
| 11:30 – 11:40 | ANALYSIS OF THE TRANSCRIPTOME OF SORAFENIB-RESISTANT HEPATOCELLULAR CARCINOMA CELLS PROVIDES INSIGHT INTO OVERCOMING THERAPEUTIC RESISTANCE Dr Tasnuva Kabir <i>Harry Perkins Institute of Medical Research, Australia</i> |  |
| 11:40 – 11:50 | AN APPROACH TO PRIORITIZING NOVEL DISEASE GENE CANDIDATES AND INVESTIGATING THEIR ROLE IN MUSCLE BIOLOGY AND DISEASE Ms Hayley Goullee <i>Harry Perkins Institute of Medical Research, Australia</i> |  |
| 11:50 – 12:50 | BREAK | |
| 12:50 – 14:20 | SESSION 3 <i>Chair: Dr Eric Alves</i> | Channel Sponsor  |
| 12:50 – 13:20 | Mentoring Speaker A/Prof Alicia Oshlack <i>Murdoch Children's Research Institute, Australia</i> | |
| 13:20 – 13:30 | VARIANTSPARK: GENERATING POLYGENIC RISK SCORES BY INCORPORATING HIGHER-ORDER EPISTASIS INTERACTIONS Dr Arash Bayat <i>CSIRO, Australia</i> |  |
| 13:30 – 13:40 | UNDERSTANDING FUNCTIONAL DRIVERS OF LIVER METASTASIS IN UVEAL MELANOMA Dr Weitao Lin <i>Edith Cowan University, Australia</i> |  |
| 13:40 – 13:50 | GENETIC DIAGNOSIS OF COMPLEX ATAXIA PEDIGREES USING SHORT-READ SEQUENCING DATA Dr Haloom Rafahi <i>The Walter and Eliza Hall Institute of Medical Research, Australia</i> |  |



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| 13:50 – 14:00 | THE USE OF CIRCULATING TUMOUR DNA (CTDNA) FOR MOLECULAR PROFILING IN PATIENTS WITH OVARIAN CANCER Dr Leslie Calapre <i>Edith Cowan University, Australia</i> | |
| 14:00 – 14:10 | STRUCTURAL VARIANT IN STMN2 IS ASSOCIATED WITH SALS DISEASE RISK, AGE OF ONSET AND CLINICAL PRESENTATION. Miss Frances Theunissen <i>The Perron Institute for Neurological and Translational Science, Australia</i> | |
| 14:10 – 14:20 | CELL TYPE AND CORTEX-SPECIFIC RNA EDITING IN SINGLE HUMAN NEURONS INFORMS NEUROPSYCHIATRIC DISORDERS Dr Brendan Ansell <i>Walter & Eliza Hall Institute of Medical Research, Australia</i> | |
| 14:20 – 14:50 | SHORT BREAK | |
| 14:50 – 15:50 | SESSION 4 <i>Chair: Mx Jennifer Currenti</i> | Channel Sponsor |
| 14:50 – 15:00 | 'A SPITTING IMAGE' OF HUMAN CYTOMEGALOVIRUS MICRO RNA MOLECULES IN CLINICAL SAMPLES Ms Shelley Waters <i>Curtin University, Australia</i> | |
| 15:00 – 15:10 | DEEP LEARNING MODEL FOR AUTOMATED CANCER CLASSIFICATION BASED ON IMAGE DATA AND MOLECULAR MARKERS Mr Xiao Tan <i>Institute for Molecular Bioscience, The University of Queensland, Australia</i> | |
| 15:10 – 15:20 | AN ASSOCIATION STUDY OF HLA-G REGULATORY REGION POLYMORPHISMS WITH RECURRENT PREGNANCY LOSS (RPL) Ms Binata Marik <i>All India Institute of Medical Sciences (AIIMS), New Delhi, India</i> | |
| 15:20 – 15:30 | GENOME-WIDE VARIABLY METHYLATED TUMOUR DNA REGIONS AND ASSOCIATION WITH OVERALL SURVIVAL IN INVASIVE LOBULAR BREAST CANCER Ms Medha Suman <i>The University of Melbourne, Australia</i> | |
| 15:30 – 15:40 | SETTING GLOBAL STANDARDS FOR GERMLINE GENOME EDITING Mr Yujia Zhu <i>National University of Singapore, Singapore</i> | |
| 15:40 – 15:50 | EPIGENETIC MODIFICATIONS UNDERLYING THE NEUROPROTECTIVE PROPERTIES INDUCED BY A SINGLE ULTRA-LOW DOSE OF THC Mr Guy Shapira <i>Tel Aviv University, Israel</i> | |
| 15:50 – 16:20 | BREAK | |

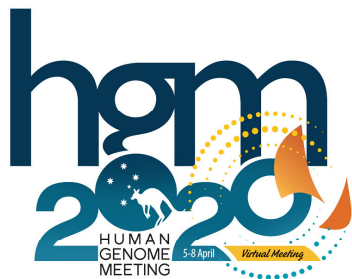


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| 16:20 – 17:00 | SESSION 5 <i>Chair: Prof David Mackey</i> | Channel Sponsor  |
| 16:20 – 17:00 | CRACKING THE CAREER CODE - AN INTERVIEW FOR ECRs Dr Myles Axton, <i>Genetics & Genomics Next, United States</i> Ms Tiffany Boughtwood, <i>Australian Genomics Health Alliance, Australia</i> Dr Ryan Taft, <i>Illumina, United States</i> Dr Kristen Nowak, <i>Office of Population Health Genomics, WA Department of Health, Australia</i> | |
| 17:00 | CLOSE | |








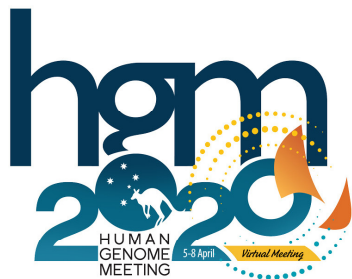
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HGM Main Meeting MONDAY 6 APRIL 2020

| | | | |
|--|---|---|---|
| 08:40 – 09:00 | | OPENING SESSION | |
| 08:40 – 08:50 | | Welcome to Country / Acknowledgment of Country | |
| 08:50 – 08:55 | | Welcome to HGM2020 Prof Alistair Forrest Convenor HGM2020 | |
| 09.00 – 11.15 | | SYMPOSIUM | |
| SYMPOSIUM 1: REGULATORY ELEMENTS 1 | | SYMPOSIUM 2: POPULATIONS PRINCIPLES AND INSIGHTS | |
| Chair: Prof Alistair Forrest | | Chair: Prof Christine Wells | |
| CHANNEL: A | | CHANNEL: B | |
| Channel Sponsor  | | Channel Sponsor  | |
| 09:00 – 09:25 MOUSE MODELS Prof Nadia Rosenthal <i>The Jackson Laboratory, United States</i> |  | 09:00 – 09:25 NATIONAL CENTRE FOR INDIGENOUS GENOMICS Simon Eastal <i>The National Centre for Indigenous Genomics, Australia</i> | |
| 09:25 - 09:50 USING hiCLIP TO DISCOVER LONG-RANGE LOOPS IN MRNAS: WHAT DO THESE LOOPS DO? Prof Nicholas Luscombe <i>The Francis Crick Institute, United Kingdom</i> |  | 09:25 - 09:50 INDONESIAN POPULATION GENETICS Prof Herawati Sudoyo <i>Eijkman Institute, Indonesia</i> | |
| 09:50 – 10:15 THE ROLE OF NONCODING REGIONS IN THE GENETIC PREDISPOSITION TO BREAST CANCER. A/Prof Juliet French <i>QIMR Berghofer Medical Research Institute, Australia</i> |  | 09:50 – 10:05 GENETIQS: DEVELOPING GUIDELINES FOR THE CONDUCT OF GENOMIC RESEARCH INVOLVING ABORIGINAL AND/OR TORRES STRAIT ISLANDER QUEENSLANDERS Mr Gregory Pratt <i>QIMR Berghofer, Australia</i> |  |
| 10:15 – 10:30 CANDIDATE SILENCER ELEMENTS IN THE HUMAN GENOME Dr David Hawkins <i>University of Washington School of Medicine, United States</i> |  | 10:05 – 10:20 MITOGENOMES REVEAL RAPID SETTLEMENT OF SAHUL AND SUBSEQUENT MOVEMENTS BETWEEN WALLACEA AND NEW GUINEA, BUT NOT AUSTRALIA Mr Gludhug Ariyo Purnomo <i>Australian Centre for Ancient DNA, The University of Adelaide, Australia</i> |  |

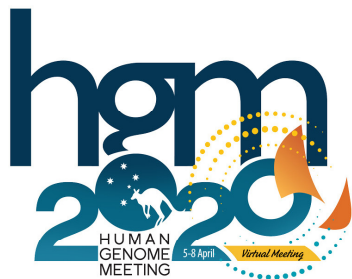


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| 10:30 – 10:45 MAXHIC: A ROBUST BACKGROUND CORRECTION MODEL TO IDENTIFY STATISTICALLY SIGNIFICANT INTERACTIONS IN HI-C AND CAPTURE HI-C EXPERIMENTS Dr Hamid Alinejad Rokny UNSW, Australia | | 10:20 – 10:35 GENETIC DRIVERS OF GENE EXPRESSION AND DNA METHYLATION ACROSS THE INDONESIAN ARCHIPELAGO Dr Irene Gallego Romero University of Melbourne, Australia | |
| 10:45 – 11:00 ALTERED 3D GENOME ORGANIZATION AND ACTIVATION OF THE MEIS1/HOXA9/PBX3 ONCOGENIC SIGNALING AXIS IN MYELOID LEUKEMIA Dr Melissa Fullwood Nanyang Technological University, Singapore | | 10:35 – 10:50 A COMPREHENSIVE SURVEY OF HUMAN ALU REPEAT DIVERSITY Mr Renzo Balboa National Centre for Indigenous Genomics, Australian National University, Australia | |
| 11:00 – 11:15 COMPLEXITY OF REGULATORY ELEMENTS EXPRESSION IN HUMAN AND PRIMATE MUSCLES REVEALED BY CAGE ANALYSIS Dr Oleg Gusev Riken, Japan | | | |
| 11:15 – 12:20 | BREAK | | |
| 12:20 – 14:50 | SYMPOSIUM | | |
| SYMPOSIUM 3: REGULATORY ELEMENTS 2 | | SYMPOSIUM 4: GENOMIC THERAPIES | |
| Chair: Dr Piero Carninci | | Chair: Dr Gina Ravenscroft | |
| CHANNEL: A | | CHANNEL: B | |
| Channel Sponsor | | Channel Sponsor | |
| 12:20 -12:45 LEARNING TO FORGET: A NEW HUMAN REPROGRAMMING STRATEGY TO ERASE SOMATIC CELL EPIGENETIC MEMORY Prof Ryan Lister University of Western Australia, Australia | | 12:20 -12:45 ANTISENSE STARTS MAKING SENSE IN DUCHENNE MUSCULAR DYSTROPHY: AN OVERVIEW Prof Steve Wilton Murdoch University, Australia | |
| 12:45 -13:10 ESCAPEE RETROTRANSPOSONS IN EMBRYONIC AND NEURONAL GENOMES Dr Seth Cheetham University of Queensland, Australia | | 12:45 -13:10 CELL-PENETRATING PEPTIDE DELIVERY OF ANTISENSE OLIGOMERS THAT MEDIATE INDIRECT MODULATION OF PRPF31 EXPRESSION TO TREAT RETINITIS PIGMENTOSA 11 Prof Sue Fletcher Murdoch University, Australia | |
| 13:10 – 13:25 GBA IN PARKINSON'S DISEASE: A REGULATORY PERSPECTIVE Ms Sophie Farrow Liggins Institute, New Zealand | | 13:10 -13:25 ALLELE-SPECIFIC CRISPR DELETION AS A THERAPEUTIC STRATEGY FOR DOMINANT SKELETAL MUSCLE A-ACTIN DISEASE Dr Rhonda Taylor Harry Perkins Institute of Medical Research, Australia | |

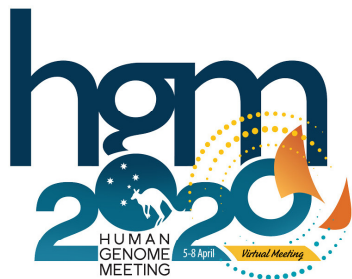


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| 13:25 – 13:40 | FUNCTIONAL INVESTIGATION INTO THE REGULATORY ROLES OF GENETIC VARIANTS OF THE BONE MORPHOGENETIC PROTEIN 4 (BMP4) GENE ASSOCIATED WITH HIGH MYOPIA IN CHINESE POPULATION Ms Regina Kwarteng <i>The Hong Kong Polytechnic University, Hong Kong1</i> |  | 13:25 – 13:40 ETHICAL REQUIREMENTS FOR HUMAN GERMLINE GENE EDITING Mr Markus Labude <i>National University of Singapore, Singapore</i> |  |
| 13:40 – 13:55 | DISCOVERY OF NEW PROMOTERS AND ENHANCERS ACTIVE IN HUMAN HEART Mr Ruslan Deviatiiarov <i>Kazan Federal University, Russian Federation</i> |  | 13:40 – 13:55 TOWARD A GENE THERAPY FOR MEN WITH PARKINSON'S DISEASE Dr Yuan Chen <i>Hudson Institute of Medical Research, Australia</i> |  |
| 13:55 – 14:10 | CORRECTION FOR BACKGROUND ALLELIC DOSAGE HIGHLIGHTS WIDESPREAD ALLELE-SPECIFIC TRANSCRIPTION FACTOR BINDING AND SUGGESTS CAUSAL VARIANTS FOR PHENOTYPIC TRAITS Prof Vsevolod Makeev <i>Vavilov Institute of General Genetics, Russian Academy of Sciences, Russian Federation</i> | | 13:55 – 14:10 TRANSLATING GENE NETWORKS INTO TREATMENTS FOR ALZHEIMER'S DISEASE Prof Eske Derks <i>QIMR Berghofer, Australia</i> |  |
| 14:10 – 14:25 | EPIGENOMICS OF THE AUDITORY SYSTEM: IMPLICATIONS FOR DEAFNESS Prof Karen Avraham <i>Tel Aviv University, Israel</i> |  | 14:10 – 14:25 ANALYSES OF 299 PHARMACOGENES IN SICKLE CELL DISEASE PATIENTS IN AFRICA Ms Khuthala Mnika |  |
| 14:25 -14:50 | EPIGENETIC MECHANISMS IN INNATE IMMUNE MEMORY Prof Musa Mhlanga <i>University of Cape Town (UCT), South Africa</i> |  | 14:25 -14:40 TARGETING mRNA EXPORT AS A NOVEL MECHANISM FOR CANCER CELL KILLING Dr Ewa Michalak <i>Peter MacCallum Cancer Centre, Australia</i> | |
| 14:50 – 15:10 | BREAK | | | |
| 15:10 – 15:55 | PLENARY SESSION 1 Chair: Prof Alistair Forrest | | Channel Sponsor  | |
| 15:30 – 15:55 | THE IMPACT OF ADMIXTURE BETWEEN MODERN AND ARCHAIC HUMANS Dr Janet Kelso <i>Max Planck Institute for Evolutionary Anthropology</i> | | | |
| 15:55 – 16:00 | BREAK | | | |

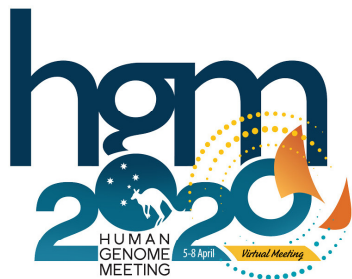


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| 16:00 – 18:00 | | SYMPOSIUM | |
|---|---------------------------|--|--|
| SYMPOSIUM 5: RARE DISEASE 1 | | SYMPOSIUM 6: NON-CODING RNAs | |
| CHAIR: Prof Jozef Gecz | | CHAIR: A/Prof Archa Fox | |
| CHANNEL: A | | CHANNEL: B | |
| Channel Sponsor | | Channel Sponsor | |
| 16:00 – 16:25 gnomAD Prof Daniel MacArthur <i>Broad Institute of MITA and Harvard, United States</i> | | 16:00 – 16:25 DRIVING IN THE DARK: MUTATED LONG NONCODING RNAs IN CANCER Prof Rory Johnson <i>University of Bern, Switzerland</i> | |
| 16:25 – 16:50 MITOCHONDRIAL DISEASES Prof Aleksandra Filipovska <i>University of Western Australia, Australia</i> | | 16:25 – 16:50 SINEUPS Mr Stefano Gustincich <i>Technologies for Life Science (LifeTech), Italy</i> | |
| 16:50 – 17:15 SICKLE CELL AND GENETICS IN TANZANIA Dr Julie Makani <i>Muhimbili University of Health and Allied Sciences</i> | | 16:50 – 17:05 GENOME-WIDE FUNCTIONAL ANNOTATION OF LONG NONCODING RNAs Dr Yulia Medvedeva <i>Institute of Bioengineering, FSI FRC Fundamentals of Biotechnology, Russian Academy of Sciences, Russian Federation</i> | |
| 17:15 – 17:30 LYFE LANGUAGES + PILBARA FACES: INDIGENOUS AUSTRALIAN 3D FACIAL VARIATION STUDY TO PROVIDE DIAGNOSIS FOR RARE AND GENETIC DISEASES - COMMUNICATION THROUGH INDIGENOUS LANGUAGES. Dr Yarlalu Thomas | | 17:05 – 17:20 THE LANDSCAPE OF CIRCULAR RNA EXPRESSION IN THE HUMAN BRAIN Dr Akira Gokool <i>School of Biotechnology and Biomolecular Science, UNSW, Australia</i> | |
| 17:30 – 17:45 MOLECULAR CHARACTERIZATION OF OCULOAUICULOVERTEBRAL SPECTRUM IN A GROUP OF EGYPTIAN PATIENTS Mrs Amira Nabil Khalaf <i>Medical Research Institute, Alexandria University, Egypt</i> | | 17:20 – 17:35 EXPANDING LNCRNA STUDIES WITH PUBLIC DOMAIN DATA THROUGH FC-R2 Dr Eddie Luidy Imada <i>Johns Hopkins University, United States</i> | |
| 17:45 – 18:00 GENETIC AND CELLULAR CHARACTERISATION OF FOCAL BRAIN MALFORMATIONS USING PATIENT-DERIVED BRAIN TISSUE Mr Wei Shern Lee <i>Murdoch Children's Research Institute, Australia</i> | | | |
| 18:00 – 20:00 | Virtual Welcome Reception | | |
| 20:00 | DAY 1 END | | |



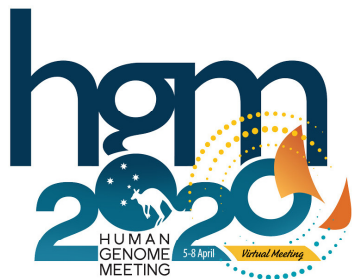
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TUESDAY 7 APRIL 2020

| | | | |
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| 07:30 – 08.45 | Oxford Nanopore Seminar | | Channel Sponsor |
| | | | |
| 07:30 - 07:45 | INTRODUCTION AND UPDATE Mr Warren Bach <i>Oxford Nanopore Technologies</i> | | |
| 07:45 – 08:05 | Technical Update Dr Michael Yarski | | |
| 08:05 – 08:25 | Mike Clark | | |
| 08:25 – 08:44 | DIRECT RNA SEQUENCING AND EARLY EVOLUTION OF SARS-CoV-2 Dr George Tairaoa | | |
| 08:44 – 08.45 | CLOSING REMARKS Dr Mari Miyamoto <i>Oxford Nanopore Technology</i> | | |
| 08:45 – 09:30 | PLENARY SESSION 2 <i>Chair: Prof Jozef Gecz</i> | | Channel Sponsor |
| | | | Agilent Trusted Answers |
| 08.45 – 09.30 | NEUROGENETICS, PSYCHENCODE Prof Daniel Geschwind <i>University of California, Los Angeles, United States</i> | | |
| 09:30 – 10:00 | BREAK | | |
| 10:00 – 12:15 | SYMPOSIUM | | |
| | SYMPOSIUM 7: SINGLE CELL | | SYMPOSIUM 8: GENOMICS FOR GOOD SESSION |
| | CHAIR: Prof Ryan Lister | | CHAIR: |
| | CHANNEL: A | | CHANNEL: B |
| | Channel Sponsor 10x GENOMICS <small>Contributed by millennium science</small> | | Channel Sponsor illumina® |
| 10:00 – 10:25 | COMPARATIVE ANALYSIS OF DISEASE-ORIENTED SINGLE-CELL TRANSCRIPTIONAL DATASET COLLECTIONS A/Prof Peter Kharchenko <i>Harvard Medical School, United States</i> | | 10.00 – 10.30 RYAN TAFT ILLUMINA |
| 10:25 – 10:40 | CELL TYPE SPECIFICITY OF INTRALOCUS INTERACTIONS REVEALS OLIGODENDROCYTE INTRINSIC MECHANISMS FOR MULTIPLE SCLEROSIS Dr Olivia Corradin <i>Whitehead Institute for Biomedical Research, United States</i> | | 10.30 – 11.00 Prof Stephen Damiani |

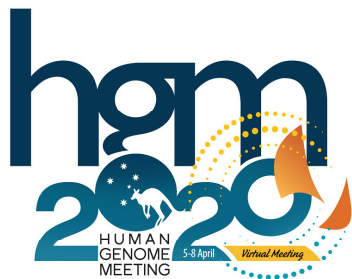


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| 10:40 – 10:55 SYSTEMATIC ASSESSMENT OF TISSUE DISSOCIATION AND STORAGE BIASES IN SINGLE-CELL AND SINGLE-NUCLEUS RNA-SEQ WORKFLOWS Dr Elena Denisenko <i>Harry Perkins Institute of Medical Research, Australia</i> | | 11:00 – 11:30 Prof Nigel Laing AO | |
| 10:55 – 11:10 SPATIAL UNDERSTANDING OF DISEASE IN SINGLE CELLS AND INTACT TISSUE SECTIONS THROUGH MACHINE-LEARNING INTEGRATION OF IMAGING AND SEQUENCING DATA. Dr Quan Nguyen <i>Institute for Molecular Bioscience, University of Queensland, Australia</i> | | 11:30 – 12:00 PANEL DISCUSSION | |
| 11:10 – 11:25 CITE-SEQ FOR INTRANUCLEAR PROTEINS: MECP2 PROTEIN EXPRESSION AT SINGLE-NUCLEUS LEVEL Ms Dulce Beatriz Vargas Landin <i>The University of Western Australia, Australia</i> | | | |
| 11:25 – 11:50 THE STEMFORMATICS PORTAL: A COMMUNITY PLATFORM FOR VISUALISATION AND DISCOVERY OF MOLECULAR STATES IN STEM CELL BIOLOGY Prof Christine Wells <i>University of Melbourne, Australia</i> | | | |
| 11:50 – 12:15 THE NANOSTRING SPATIAL GENOMICS ROADMAP: CANCER TRANSCRIPTOME ATLAS TO WHOLE TRANSCRIPTOMES AND BEYOND. Joseph Beecham <i>Nanostring Technologies, United States</i> | | | |
| 12:15 – 13:00 | BREAK | | |
| 13:00– 13:45 | PLENARY SESSION 3 <i>Chair: A/Prof Elin Gray</i> | | Channel Sponsor |
| 13:00 – 13:45 | BREAST CANCER SUSCEPTIBILITY Prof Georgia Chenevix-Trench <i>QIMR-Berghofer Medical Research Institute, Australia</i> | | |



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| 13:45 – 15:45 | | SYMPOSIUM | |
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| SYMPOSIUM 9: FUNCTIONAL GENOMICS | | SYMPOSIUM 10: POPULATION GENETICS | |
| CHAIR: A/Prof Julian Heng | | CHAIR: Prof David Mackey | |
| CHANNEL: A | | CHANNEL: B | |
| Channel Sponsor | | Channel Sponsor | |
| | | | |
| 13:45 – 14:10 FUNCTIONAL GENOMICS APPROACHES TO DISSECT NEURODEGENERATIVE DISEASES Prof Peter Heutink <i>German Center for Neurodegenerative Diseases (DZNE)-Tübingen, Germany</i> | | 13:45 – 14:10 SINGLE CELL eQTLs A/Prof Joseph Powell <i>Garvan-Weizmann Centre for Cellular Genomics, Australia</i> | |
| 14:10 – 14:35 HUMAN STEM CELL BASED FUNCTIONAL NEUROGENOMICS Prof Ernst Wolvetang <i>Australian Institute for Bioengineering and Nanotechnology, University of Queensland, Australia</i> | | 14:10 – 14:25 THE SEARCH FOR GENES INFLUENCING THE STRUCTURE OF THE HUMAN CORTEX Dr Jodie Painter <i>QIMR Berghofer Medical Research Institute, Australia</i> | |
| 14:35 – 14:50 LOOKING BEYOND THE EXOME: INVESTIGATING THE ROLE OF STRUCTURAL VARIATION IN THE PATHOGENIC LANDSCAPE OF HEREDITARY NEUROPATHIES Prof Marina Kennerson <i>ANZAC Research Institute, Australia</i> | | 14:25 – 14:40 WHERE IN THE HUMAN GENOME DOES COMPLEX TRAIT HERITABILITY LIE? David Balding <i>University of Melbourne, Australia</i> | |
| 14:50 – 15:05 AUTOMATIC IDENTIFICATION OF SMALL MOLECULES THAT PROMOTE CELL CONVERSION Dr Erik Arner <i>RIKEN, Japan</i> | | 14:40 – 14:55 IMPACT OF STRUCTURAL VARIATIONS ON HUMAN GENOME TOPOLOGY AT THE POPULATION SCALE Prof Dariusz Plewczynski <i>University of Warsaw, Poland</i> | |
| 15:05 – 15:20 ARE GENES ENCODING CYLD INTERACTORS IMPLICATED IN FRONTOTEMPORAL DEMENTIA AND AMYOTROPHIC LATERAL SCLEROSIS? Dr Carol Dobson-Stone <i>University of Sydney, Australia</i> | | 14:55 – 15:10 ROLE OF TESTOSTERONE IN THE SEX-SPECIFIC GENETIC ARCHITECTURE OF COMPLEX TRAITS Dr Allan McRae <i>Institute for Molecular Bioscience, The University of Queensland, Australia</i> | |
| | | 15:10 – 15:25 GWAS META-ANALYSIS OF MOTHERS OF DIZYGOTIC TWINS PLUS CONTROLS REVEALS 15 LOCI CONTROLLING FEMALE REPRODUCTION Prof Nick Martin <i>QIMR, Australia</i> | |
| 15:25 – 15:50 | BREAK | | |

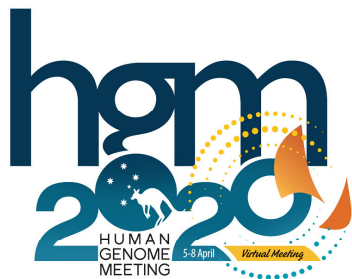


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| 15.50 – 17:40 | | SYMPOSIUM | |
|---|---|---|---|
| SYMPOSIUM 11: RARE DISEASE 2 | | SYMPOSIUM 12: CANCER GENOMICS 1 | |
| CHAIR: Prof Karen Avraham | | CHAIR: A/Prof Elin Gray | |
| CHANNEL: A | | CHANNEL: B | |
| Channel Sponsor | | Channel Sponsor | |
|  | |  | |
| 15:50 – 16:15 | GUARDIAN (RARE DISEASE) Prof Vinod Scaria <i>CSIR Institute of Genomics and Integrative Biology</i> |  | 15:50 – 16:15 FINDING CLINICALLY RELEVANT VARIANTS USING CANCER TRANSCRIPTOMES A/Prof Alicia Oshlack <i>Murdoch Children's Research Institute, Australia</i> |
| 16:15 – 16:40 | HUMAN IMPRINTING DISORDERS –PRACTICE, PROBLEMS AND PROGRESS Prof Karen Temple <i>University of Southampton; Wessex Genome Medicine Centre, United Kingdom</i> | | 16:15 – 16:40 GENOME-WIDE ANALYSIS OF ctDNA FOR CANCER MONITORING AND STRATIFICATION Dr Ellen Heitzer <i>Institute of Human Genetics, Medical University Graz, Austria</i> |
| 16:40 – 16:55 | IN SEARCH OF GENETIC MARKERS FOR NON-SYNDROMIC HEARING IMPAIRMENT AND DEVELOPMENT OF EFFECTIVE AND AFFORDABLE TOOL FOR TESTING A DEAFNESS GENE IN GHANA Mr Samuel Adadey <i>University of Ghana, Ghana</i> |  | 16:40 – 16:55 IMPACT OF GERMLINE COPY NUMBER VARIATION ON ENDOMETRIAL CANCER RISK Ms Cassie Stylianou <i>University of Otago, Christchurch, New Zealand</i> |
| 16:55 – 17:10 | CONFIRMATION OF GENETIC DIVERSITY OF AFRICAN POPULATION: REPORTS FROM CHARCOT-MARIE-TOOTH DISEASE STUDY IN MALI Dr Abdoulaye Yalcouyé |  | 16:55 – 17:10 WHOLE-GENOME CHARACTERISATION OF ENDOMETRIAL CANCER PATIENT-DERIVED XENOGRAFTS Dr Olga Kondrashova <i>QIMR Berghofer Medical Research Institute, Australia</i> |
| 17:10 – 17:25 | FANTOM5 GENE EXPRESSION DATA INFORMS THE IDENTIFICATION OF TWO NOVEL HUMAN DISEASE GENES Dr Gina Ravenscroft <i>Harry Perkins Institute, Australia</i> |  | 17:10 – 17:25 SINGLE-CELL TRANSCRIPTOME REVEALS MAMMARY EPITHELIAL LINEAGE RELATIONSHIPS UNDER NORMAL AND TUMOUR MICROENVIRONMENT Dr Bhupinder Pal <i>Olivia Newton-John Cancer Research Institute, Australia</i> |
| 17:25 – 17:40 | A MĀORI SPECIFIC RFC1 PATHOGENIC REPEAT CONFIGURATION IN CANVAS, LIKELY DUE TO A FOUNDER ALLELE Ms Sarah Beecroft <i>Harry Perkins Institute, Australia</i> |  | 17:25 – 17:40 USING PAIRWISE BIOLOGICAL RELATIONS BETWEEN FEATURES TO REDUCE OVER-FITTING IN PRECISION MEDICINE Prof Luigi Marchionni <i>Johns Hopkins University, United States</i> |
| 17:40 | DAY 2 END | | |



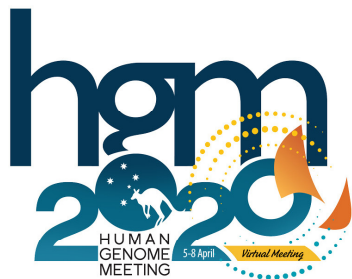
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WEDNESDAY 8 APRIL 2020

| | | | |
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| 07:30 – 08:45 | MGI Workshop | | |
| 08:45 – 09:30 | PLENARY SESSION 4 <i>Chair: Dr Nic Waddell</i> | | Channel Sponsor |
| 08:45 – 09:30 | PRECISION MEDICINE IN THE CANCER CLINIC Dr Razelle Kurzrock <i>University of California San Diego Moores Cancer Center, United States</i> | | |
| 09:30 – 10:00 | BREAK | | |
| 10:00 – 12:00 | SYMPOSIUM | | |
| | SYMPOSIUM 13: CANCER GENOMICS 2 | | SYMPOSIUM 14: CLINICAL GENETICS |
| | CHAIR: Dr Raelene Endersby | | CHAIR: Prof Ingrid Winship |
| | CHANNEL: A | | CHANNEL: B |
| | Channel Sponsor | | Channel Sponsor |
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| 10:00 – 10:25 | CIRCULATING CELL FREE GENOME ATLAS Prof Minetta Liu <i>Mayo Clinic, United States</i> | | 10:00 – 10:25 THE UNDIAGNOSED SECOND DIAGNOSIS Dr Madhuri Hegde <i>Georgia Institute of Technology, Global Laboratory Services, PerkinElmer Inc., United States</i> |
| 10:25 – 10:50 | DECODING PANCREATIC CANCER Prof Sean Grimmond <i>University of Melbourne Centre for Cancer Research, Australia</i> | | 10:25 – 10:50 NEW MOLECULAR APPROACHES TO THE DIAGNOSIS OF RARE GENETIC DISORDERS Dr Carlos Bacino <i>Texas Children's Hospital, United States</i> |
| 10:50 – 11:15 | BIO-RAD SPONSORED TALK Dr Elin Gray <i>Edith Cowan University, Australia</i> | | 10:50 – 11:15 LOW-PASS GENOME SEQUENCING EXPLORES COMPLEXITY OF CHROMOSOMAL ABNORMALITIES IN RECURRENT MISCARRIAGE COUPLES Dr Richard Choy <i>The Chinese University of Hong Kong, Hong Kong</i> |
| 11:15 – 11:30 | SINGLE CELL ATLAS OF HUMAN HEPATOCELLULAR CARCINOMA Dr Ankur Sharma <i>Genome Institute of Singapore, Singapore</i> | | 11:15 – 11:30 PHENOTYPE MATTERS WHEN PRIORITISING VARIANTS Mrs Denise Anderson <i>Telethon Kids Institute, Australia</i> |

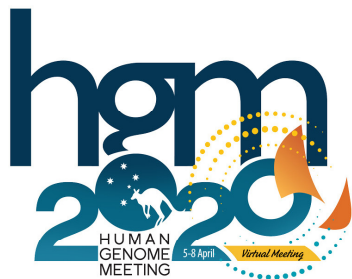


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| 11:30 – 11:45 CHROMATIN INTERACTOME MAPPING IDENTIFIES TARGET GENES AT 139 INDEPENDENT BREAST CANCER RISK SIGNALS A/Prof Stacey Edwards <i>QIMR Berghofer Medical Research Institute, Australia</i> | | 11:30 – 11:45 A LINKED-READ GENOME SEQUENCING APPROACH EXPANDS THE GENETIC CONTRIBUTORS TO MEIER-GORLIN SYNDROME Dr Louise Bicknell <i>University of Otago, New Zealand</i> | |
| 11:45 – 12:00 DEVELOPING PERSONALIZED CHEMOTHERAPY-TARGETED TREATMENT FOR SARCOMA PATIENTS VIA PDX TESTING AND PATIENT/TUMOUR/PDX GENOMIC ANALYSIS. A/Prof Evan Ingley <i>Harry Perkins Institute of Medical Research, Australia</i> | | 11:45 – 12:00 AN ARTIFICIAL INTELLIGENCE METHOD FOR FAST DIAGNOSIS OF RARE GENETIC DISEASE Dr Francisco De La Vega <i>Fabric Genomics, United States</i> | |
| 12:00 – 13:00 | BREAK | | |
| 13:00 – 15:15 | SYMPOSIUM | | |
| SYMPOSIUM 15: IMMUNOGENOMICS | | SYMPOSIUM 16: NEW FRONTIERS | |
| CHAIR: | | CHAIR: Dr Christian Pflueger | |
| CHANNEL: A | | CHANNEL: B | |
| Channel Sponsor | | Channel Sponsor | |
| | | | |
| 13:00 – 13:25 IMMUNOGENETICS OF HLA AND NK RECEPTORS Prof John Trowsdale | | 13:00 – 13:25 THE NEW ERA OF TELOMERE-TO-TELOMERE GENOMICS: EXPANDING STUDIES OF HIDDEN GENETIC VARIATION AND FUNCTION IN THE UNCHARTED REGIONS OF THE HUMAN GENOME Dr Karen Miga <i>University of California Santa Cruz Genomics Institute, United States</i> | |
| 13:25 – 13:50 A/Prof Silvana Gaudieri <i>Murdoch University, Australia</i> | | 13:25 – 13:50 BIG DATA AND HEALTH Prof Michael Snyder <i>Stanford University - Genetics, United States</i> | |
| 13:50 – 14:15 STRUCTURAL INSIGHTS INTO IMMUNE RECOGNITION Dr Julian Vivian <i>Monash University, Australia</i> | | 13:50 – 14:05 ENABLING SINGLE MOLECULE TRANSCRIPTOMICS AT POPULATION SCALE Prof Eduardo Eyras <i>Australian National University, Australia</i> | |
| 14:15 – 14:30 CONCORDANCE HLA TYPING REVEALS NOVEL PROTECTIVE VARIANTS IN HLA-B*27 POSITIVE ANKYLOSING SPONDYLITIS Dr Jennifer Meadows <i>Uppsala University, Sweden</i> | | 14:05 – 14:20 A MACHINE LEARNING APPROACH TO REDUCE FALSE POSITIVES IN IDENTIFYING STRUCTURAL VARIATIONS Ms Hansol Park <i>Korea Advanced Institute of Science and Technology (KAIST), South Korea</i> | |



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| 14:30 – 14:45 | MULTIDIMENSIONAL SINGLE-CELL PLATFORMS DEFINE DISTINCT GENETIC, CELLULAR AND MOLECULAR PROFILES AT THE SITE OF TISSUE DAMAGE IN STEVENS-JOHNSON SYNDROME AND TOXIC EPIDERMAL NECROLYSIS. Dr Andrew Gibson <i>Institute of Immunology and Infectious Disease (IID), Murdoch University, Australia</i> | | 14:20 – 14:35 CLINICAL VARIANT ANNOTATION WITH MULTIPLEXED ASSAYS OF VARIANT EFFECT Dr Alan Rubin <i>The Walter and Eliza Hall Institute of Medical Research, Australia</i> | |
| 14:45 – 15:00 | THE BURNING QUESTION: DO TNF-BLOCK GENOTYPES INFLUENCE SUSCEPTIBILITY TO HIV-ASSOCIATED SENSORY NEUROPATHY IN INDONESIA AND SOUTH AFRICANS? Ms Jessica Gaff <i>Curtin Health Innovation Research Institute, Australia</i> | | 14:35 – 14:50 ENGINEERING OF AN EFFICIENT AND SPECIFIC PLATFORM FOR PROGRAMMABLE RNA BASE EDITING Dr Meng How Tan <i>Nanyang Technological University, Singapore</i> | |
| | | | 14:50 – 15:15 OXFORD NANOPORE Dr Clive Brown | |
| 15:15 – 15:30 | BREAK | | | |
| 15:30 – 16:45 | CLOSING SESSION <i>Chair: Prof Alistair Forrest</i> | | | |
| 15:30 – 16:00 | Acknowledgements and prizes | | | |
| 16:00 – 16:45 | Closing Remarks and HUGO HGM 2021 Announcement | | | |
| 16:45 | MEETING CONCLUDES | | | |