

Wednesday, 22 February 2023

| Time | Auditorium 1 |
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| 09h00 - 16h00 | H3A Bionet: Celebrating 10 Years of Advancing Bioinformatics Research and Capacity Building in Africa (Hall 8, CTICC 2) |
| 09h00 - 14h00 | Pre-congress workshop: Genomics in Paediatric Care (hosted by Illumina) (Meeting Block 2.60, Level 2) |
| 09h00 - 13h00 | IFHGS EXCO Meeting (Meeting Block 2.44, Level 2) |
| 12h00 - 19h00 | Registration Open (Registration Foyer, Ground Floor) |
| | <p>Opening Ceremony (Ballroom, Level 1)</p> <p>Chair: Raj Ramesar</p> <p>16h30 - 16h50 Welcome Address (<i>Raj Ramesar & Charles Rotimi, Co-Chairs of the Local Organising Committee</i>)</p> <p>16h50 - 16h55 Welcome from the President of the African Society of Human Genetics (<i>Ambroise Wonkam</i>)</p> <p>16h55 - 17h00 Welcome from the President of the Southern African Society of Human Genetics (<i>Collet Dandara</i>)</p> <p>17h00 - 17h10 Welcome from IFHGS President and Programme Introduction (<i>Michele Ramsay, Chair of International Scientific Programme Committee</i>)</p> <p>17h10 - 17h20 Naledi Pandor (South African Minister of International Relations and Cooperation)</p> <p>17h20 - 18h20 Opening Address: "The Rise of Genomics in Africa" (<i>Francis Collins, Former director, National Institutes of Health, USA</i>)</p> |
| 16:30 - 18:30 | 18h20 - 18h30 Acknowledgements & Declare Congress Open (<i>Raj Ramesar & Charles Rotimi</i>) |
| 18:30 - 20:00 | Welcome Reception and Official Opening of Exhibition (Exhibition Hall 1 & 2) |

Thursday, 23 February 2023

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| 07:30 - 17:00 | Registration (Registration Foyer - Ground Level) | | | | | | | | |
| 09:00 - 16:00 | Exhibition (Hall 1 & 2 - Ground Level) | | | | | | | | |
| 07:30 - 08:45 | Meet the Professor | | | | | | | | |
| | Plenary 1 - Genomics and anthropology - insights on the peopling of Africa Venue: Ballroom (Level 1) Chairs: Michele Ramsay and Charles Rotimi | | | | | | | | |
| 09:00 - 10:30 | Chairs: Michele Ramsay and Charles Rotimi | | | | | | | | |
| 09:00 - 09:45 | "The genetic history of Africa based on modern and ancient DNA " - Carina Schlebusch, Sweden | | | | | | | | |
| 09:45 - 10:30 | "Academic and Social Consequences of Past Classifications of African Peoples" - Shomarka Keita, USA | | | | | | | | |
| 10:30 - 11:00 | Tea / Coffee Break | | | | | | | | |
| 11:00 - 12:30 | INVITED SESSIONS | Session 1 - Genetic and Epigenetic Links Between Early Life and Later Life Health Outcomes Chair: Fasil Tekola-Ayele, USA (Venue: Ballroom East, Level 1) | Session 2 - Global Perspectives on Return of Individual Research Results to Participants Chair: Johanna Maria Catharina Blom, Italy (Venue: Meeting Block 1,61 - 1,62 Level 2) | Session 3 - Hemoglobinopathies: Challenges and Perspectives Chair: Martine de Rycke, Belgium (Venue: Meeting Block 1.41 - 1,42: Level 1) | Session 4 - Vascular Anomalies: from Somatic Mutations towards Targeted Therapies Chair: Miikka Vikkula, Belgium (Venue: Meeting Block 1.43 - 1,44: Level 1) | Session 5 - The Contribution of African Genomics Research to Innovation in Psychiatry Chairs: Shareefa Dalvie, South Africa & Nastassja Koen, South Africa (Venue: Meeting Block 1,63-1,64, Level 1) | Session 6 -The future of Genomics (ClinGen and ClinVar) Chair: Erin Rooney Riggs, USA (Venue: Meeting Block 2.6, Level 2) | Session 7 - Cancer Genetics Chair: Raj Ramesar, South Africa (Venue: Meeting Block 2.44-2.46, Level 2) | Session 8 - Susceptibility to Infectious Disease - from genetics to exposome Chairs: Inga Prokopenko, UK & Brent Richards, Canada (Venue: Meeting Block 2.41 - 2.43, Level 2) |
| | | Genetic and epigenetic regulation of fetal growth and links with adult cardiometabolic diseases: Fasil Tekola-Ayele, USA | Return of Results in a Healthy Cohort: The CHRIS Policy: Deborah Mascalzoni, Sweden/Italy | Perspectives in Genomics and Sickle Cell Disease Care in Africa: Ambroise Wonkam, USA/South Africa | Vascular Malformations: Genetic Discoveries on Pathophysiology: Miikka Vikkula, Belgium | An overview of NeuroGAP, including the rationale for launching the initiative and primary findings from this large-scale African genomic study: Lukoye Atwoli, Kenya | Panel: Nadia Carstens, South Africa; Melissa Nel, South Africa; Joao Bosco de Oliveira Filho, Brazil (pre-recorded) | GWAS of Cancers in African Populations: Chris Mathew, South Africa/UK | Understanding how obesity causes severe COVID-19: Brent Richards, Canada |
| | | Genetic links between birthweight and type 2 diabetes in adulthood:Rachel Freathy, USA | Ethical Framework of the Hong Kong Genome Project on Return of Individual Research Results as Benefit Sharing: Calvin Ho, China | Hemoglobinopathies: Molecular genetics, diagnostic applications, challenges and future perspectives: Jan Traeger-Synodinos, Greece (pre-recorded) | Vascular Malformations: Lessons Learned from Preclinical Models: Taija Mäkinen, Sweden | | ClinVar: Perspectives from Global Users/Submitters: João Bosco de Oliveira Filho, Brazil | Genomic Landscape of African Cancers: Sandeep Dave, USA | Applied genetic epidemiology and infectious disease: an emerging story: Fergus Hamilton, UK (pre-recorded) |
| | | Genetic and epigenetic modifiers and mediators of perinatal environmental exposures and offspring cardiovascular health: Daniel Enquobahrie, USA | Communitarian Ethics and the Return of Genetic Results in Africa: Nchangwi Syntia Munung, South Africa | Preimplantation Genetic Testing for hemoglobinopathies and HLA typing: Martine De Rycke, Belgium | Vascular Malformations: Towards Precision Therapies: Laurence M. Boon, Belgium | Population genomic resources to empower the PUMAS Project: Lerato Majara, South Africa | The Clinical Genome Resource (ClinGen): An Overview: Erin Rooney Riggs, USA | Affordable prevention of hereditary colon cancer: Sir John Burn, UK | Genetic Mechanisms of Critical Illness in Covid-19: James F Wilson, UK |
| | | Using electronic health records to identify pleiotropy and shared genetic architectures of complex diseases: Tesfaye Mersha, USA | The FACILITATE ethical framework on the return of data to patients in clinical trials: Ciara Staunton, Italy | | | | Utilizing ClinGen Resources: Danielle Azzariti, USA | | |
| 12:30 - 14:00 | Lunch Break | | | | | | | | |
| 13:00 - 14:00 | Lunch Symposia / Workshops | Proposing a network of Genomic Centres of Excellence in Africa (GenCoE) (Ballroom East, Level 1) | Lunchtime Symposium 2 | Rare Diseases Celebration Day (Meeting Block 1.41 & 1.42, Level 1) | H3Africa AGenDA Meeting: 14h15 - 14h45, Meeting Block 1.43 & 1.44, Level 1 | Workshops / Focus Groups 3 | Workshops / Focus Groups 4 | Workshops / Focus Groups 5 | Workshops / Focus Groups 6 |
| 14:00 - 15:00 | POSTER SESSION (Venue: Exhibition Hall - Hall 1 & 2, Ground Level) | | | | | | | | |
| 15:00 - 15:30 | Tea / Coffee Break | | | | | | | | |
| | | MEETING ROOM : Ballroom East | MEETING ROOM : Room 1,61 - 1,62 | MEETING ROOM : Room 2,6 | MEETING ROOM: 1,43 - 1,44 | MEETING ROOM:1,63 -1,64 | MEETING ROOM: 2,41 - 2,43 | MEETING ROOM: 2,44 - 2,46 | MEETING ROOM: 1,41 - 1,42 |
| 15:30 - 16:00 | Session Chairs | Karen Fieggen and Tjitske Kleefstra | Gerome Breen and Magdalena Mroczek | Eimear Kenny and Noemi Piga | Georgia Chenevix-Trench and Lindie Lamola | Amy Bentley and Tinashe Chikowore | Danielle Azzariti and Syntia Munung | Aime Lumaka and Candice Feben | Cesar Augusto Fortes Lima and Dhriti Segur |
| | Invited Keynote Speakers | Keynote 1 - Mendelian Phenotypes through the lens of OMIM: Ada Hamosh, USA | Keynote 2 - The genetics of major depression in diverse populations: Karoline Kuchenbaecker, UK | Keynote 3 - Multivariate methods and their applications in complex trait genetics: Inga Prokopenko, UK | Keynote 4 - Genomes and Genomics of Breast Cancers in Asian Patients: Soo-Hwang Teo, Malaysia | Keynote 5 - Leveraging multi-omics for understanding mechanisms of hypertension: Patricia Munroe, UK | Keynote 6 - Implementing Equitable Genomic Medicine: Challenges and Opportunities: Cathy Wicklund, USA | Keynote 7 - New therapies for skeletal dysplasia (dwarfism): Ravi Savarinrayan, Australia | Keynote 8 - Connecting Population Genetics to Human Diseases: Special Referecne to Asia and the Indian Subcontinent: Analabha Basu, India |
| | Oral Presentations | | | | | | | | |
| 16:00 - 16:15 | | OP001. The utility of clinical exome sequencing as a first-tier diagnostic tool in critically ill infants in South Africa: Lisa Campbell, South Africa | OP007. Polygenic risk scores for lifespan, BMI, and chronic pain predict a medication-based Rx-Risk Comorbidity Index in the Australian Genetics of Depression Study: Penelope Lind, Australia | OP013. Multi-omics study highlights several metabolites and gene expression levels as predictors of weight loss maintenance: Tingyu Guo, UK | OP019. CRISPR screens of candidate breast cancer risk genes using multiple assays to identify genes involved in proliferation, tumorigenicity, DNA damage response and immune-surveillance: Georgia Chenevix-trench, Australia | OP025. Detailing the Inflammatory Cascade of Severe Malaria through Transcriptomic, Proteomic, and Metabolomic Analyses: Rafal Sobota, USA | OP031. Whole genome sequencing (WGS) partnership program: a risk-sharing agreement to implement WGS as a first line test in paediatric monogenic disease: Ben Lundie, Australia | OP037. Development of a polygenic score to predict cisplatin-induced ototoxicity: Deanne Nixie Miao, Canada | OP043. Core Investigation of Exonic Variations in Olfactory Receptor Genes (ORGs) in Indigenous Populations from Southeast Asia: Sze Mei Lee, Malaysia |
| 16:15 - 16:30 | | OP002. Exome Sequencing in Childhood Onset Essential Hypertension Implicates Monogenic Disruption of Vascular Integrity in Disease Pathogenesis: Edmond Wonkam Tingang, USA | OP008. Rare-variant analysis of whole-genome sequence data obtained from multi-ancestry families identifies new genes associated with late-onset Alzheimer's disease: Suzanne Leal, USA | OP014. Circulating cytokines and their role in type 2 diabetes among Africans: A Mendelian randomization study: Karlijn Meeks, USA | OP020. Targeted delivery of miR-218 via decorated hyperbranched polyamidoamine for liver cancer regression: Mahmoud Elhefnawi, Egypt | OP026. Investigating the impacts of age at menarche on cognitive function based on a Mendelian randomization approach using a nationwide cohort from the Taiwan Biobank: Meng-Che Tsai, Taiwan | OP032. What are patient perspectives on privacy and trust in digital genomic tools? A qualitative study: Vedika Jha, Canada | OP038. Large cardiac arrhythmia gene panel genetic testing as prognostic tool for Brugada syndrome: Sonia Van Dooren, Belgium | OP044. Evidence of recent positive selection in the innate immune system and ion homeostasis among populations of diverse ancestries in mainland India: Chandrika Bhattacharyya, India |
| 16:30 - 16:45 | | OP003. Comprehensive cytogenetic study of a large cohort of Egyptian referral patients with Disorders of Sex Development (DSD): Mona Mekkawy | OP009. Characterizing the shared genetic influences between schizophrenia and reaction time variability: Olivia Wootton, South Africa | OP015. GWAS meta-analysis in SCAD, a women predominant ischemic heart disease, reveals common variants and genes related to artery integrity and tissue-mediated coagulation: Takiy Berrandou, France | OP021. Mismatch Repair variant classification with ACMG/AMP criteria and comparison with Bayesian probability framework: Finlay Macrae, Australia | OP027. Acceptability of Dried Blood Spot collection by Primary Caregivers of Filipino patients with Maple Syrup Urine Disease and Phenylketonuria: A Philippine's experience on Long Term Management for Newborn Screening patients:Roxanne Janica Merencilla, Phillipines | OP033. Navigating Layers of Uncertainty: Perspectives from South African Genetic Counselling Interactions: Meagan Scott, South Africa | OP039. Developing an integrated risk score for cardiovascular diseases in African populations: Michelle Kamp, South Africa and UK | OP045. Global Proteomic Analyses of Type 2 Diabetes and Obesity in West Africans: Adebowale Adeyemo, USA |

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| 16:45 - 17:00 | | OP004. Deciphering Developmental Disorders in Africa (DDD-Africa) – Clinical lessons learnt following whole exome sequencing in an African setting: <i>Amanda Krause, South Africa</i> | OP010. Variant-to-gene mapping paired with cross-species validation reveals novel effector genes for insomnia: <i>Amber Zimmerman, USA</i> | OP016. Non-additive polygenic score: <i>Rikifumi Ota, Japan</i> | OP022. Novel biological insights into the genetic architecture of bipolar disorder form multi-ancestry genome-wide association studies: <i>Kevin O'Connell</i> | OP028. Novel Copy Number Variants associated to venous thromboembolism (VTE) and warfarin dose requirement in African Americans: <i>Carolina Clark OBO Minoli Perera, USA</i> | OP034. Engaging with Local Communities in Botswana: Understanding impact of Cultural Values, Norms and Beliefs on Gene-Editing Research: <i>Sethomo Koloi-Keaikitse</i> | OP040. Pleiotropic association of rare variation with the plasma lipidome and type 2 diabetes: <i>Joanne E Curran, United States</i> | OP046. Genome-wide association study of oesophageal cancer identifies trans-ethnic risk variants in African and Chinese populations: <i>Wenlong Chen, South Africa</i> |
| 17:00 - 17:15 | | OP005. Molecular diagnosis of RASopathy patients using a next generation sequencing multi-gene targeted panel and multiplex ligation-dependent probe amplification analysis: <i>Maria Mudau, South Africa</i> | OP011. The incorporation of novel audiogram classification strategies to identify genes and pathways involved in age-related hearing loss: <i>Samah Ahmed, Canada</i> | OP017. BridgePRS: A powerful trans-ancestry Polygenic Risk Score method: <i>Paul O'Reilly, USA</i> | OP023. Investigating the role of polygenic background in neurodevelopmental disorders: <i>Qinqin Huang</i> | OP029. Discovery of the first two genome-wide significant loci for ectopic pregnancy highlights MUC1 as a potential candidate gene and identifies genetic and phenotypic relationships across traits: <i>Natàlia Pujol Gualdo, Estonia</i> | OP035. Personal values versus professional role: healthcare professionals experiences with offering termination of pregnancy for fetal abnormalities: <i>Malebo Malope, South Africa</i> | OP041. Genetic relationships and causality between overall and central adiposity and breast, prostate, lung and colorectal cancer: <i>Jared Maina, France</i> | OP047. Evidence of potential natural selection in African American individuals post admixture: <i>James Jaworski, USA</i> |
| 17:15 - 17:30 | | OP006. Loss-of-function of AMFR causes autosomal recessive hereditary spastic paraplegia by altering lipid metabolism: <i>Stefan Barakat, Netherlands</i> | OP012. Genome-wide association study of breast density among women of African Ancestry: <i>Shefali Verma, USA</i> | OP018. Recessive effects of protein-coding variants in a cohort of 44,000 British Pakistanis and Bangladeshis: <i>Teng Hiang Heng, UK</i> | OP024. The Genetics of Extreme Birthweight and its Relationship with Cardiometabolic Disease: <i>Gunn-Helen Moen, Australia</i> | OP030. Genomic spectrum of non-syndromic hearing impairment in Senegal: <i>Rokhaya Ndiaye, Senegal</i> | OP036. Genetic risk prediction of high-risk human papillomavirus infection and cervical cancer: <i>Triin Laisk, Estonia</i> | OP042. An assessment of the copy number variation landscape in Sub-Saharan African populations: <i>Laura Cottino, South Africa</i> | Blank |
| 18:00 - 19:30 | Southern African Society of Human Genetics (SASHG) - BGM (Meeting Block 1.41 & 1.42) | | | | | | | | |
| 19:30 - late | Workshop: It's more than just training: Capacity building in context. (Hosted by Wellcome Connecting Science) (Meeting Block 1.43 & 1.44) | | | | | | | | |
| | President's Dinner (by invitation only) | | | | | | | | |

Friday, 24 February 2023

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| 07:30 - 17:00 | Registration open (entrance to Hall 1, Ground Level) | | | | | | | | |
| 09:00 - 16:00 | Exhibition open (Hall 1 & 2, Ground Level) | | | | | | | | |
| 07:30 - 08:45 | | | | | | | | | |
| 09:00 - 10:30 | Plenary 2 - Translational Genomics and Implementation Science Chairs: David Amor and Kelly Ormond Venue: Ballroom, Level 1 | | | | | | | | |
| 09:00 - 09:45 | *Non-Invasive Prenatal Testing as an Archtype of Translational Genomics* - Dennis Lo, Hong Kong (pre-recorded) | | | | | | | | |
| 09:45 - 10:30 | *Genomics and the Future of Medicine* - Mayana Zatz, Brazil (pre-recorded) | | | | | | | | |
| 10:30 - 11:00 | Tea / Coffee Break | | | | | | | | |
| 11:00 - 12:30 | Session 9 - Diversity for the Future of Precision Medicine & Health Population Screening Chair: Vicky Nembaware, South Africa (Venue: Ballroom East, Level 1) | Session 10 -Global Initiatives in Genomic Medicine Workforce Development: Ensuring Best Practice in Genomic Education Chairs: Tina-Marie Wessels, South Africa & Sylvia Metcalfe, Australia (Venue: Meeting Block 1,61 - 1,62 , Level 1) | Session 11 -Therapeutic Approaches to Genetic Disorders - Before, During and After Chairs: Jennifer Posey, USA & Careni Spencer, South Africa (Venue: Meeting Block 1.41 - 1,42, Level 1) | Session 12- Advances in Genetics and Gene Directed Therapies for Inherited Retinal Diseases Chair: Alison Hardcastle, UK (Venue: Meeting Block 1,43 - 1,44 Level1) | Session 13 - Methodology and Considerations for Analyses in Diverse Populations Chairs: Marika Kaakinen, UK & Krista Fischer, Estonia (Venue: Meeting Block 1.63 - 1.64, Level 1) | Session 14 - AI-driven next-generation phenotyping: impact on the clinical diagnostic process Chairs: Shahida Moosa, South Africa & Thomy de Ravel, Belgium (Venue: Meeting Block 2.41 - 2.43, Level 2) | Session 15 - Multi-Omics Chair: Ayse Demirkan, UK & Inga Prokopenko, UK (Venue: Meeting Block 2.44 - 2.46, Level 2) | Session 16 - Sustainable Development Goals (SDG 2030) For Genetic and Genomic Disorders Chair: Meow-Keong Thong, Malaysia (Venue: Meeting Block 2.6 Level 2) | |
| | INVITED SESSIONS | | | | | | | | |
| | Lack of Diversity Remains an Unsolved Problem for Precision Medicine but Hopefully Not for Long: Janina Jeff, USA | Ensuring Best Practice in Genomics Education and Evaluation: The Australian Genomics Workforce and Education Research Program: Amy Nisselle, Australia | Novel Therapies: Achondroplasia as a Model: Melita Irving, UK | Advances in Defining the Inherited Retinal Disease Genome: Alison Hardcastle, UK | The Power of Diverse Populations for Discovery and Translation of Genome-Wide Association Studies: Andrew Morris, UK | Face2Gene: a toolbox for AI-based dysmorphisms evaluation: Nicole Fleischer, USA | The use of multi-omics in African American Genomic Medicine: Minoli Perera, USA | Achieving Sustainable Development Goals for Congenital Disorders: Challenges and Opportunities: Meow Keong Thong, Malaysia | |
| | Novel and Known Genes associated with Cardiovascular Disease Risks in African Populations: Palwendé Boua, Burkina Faso | Supporting The Implementation of a National Genomics Medicine Service In England: Michelle Bishop, UK | Genetic Biobanks, Registries, Patient Organisations: Towards Therapies for FOP: Luca Sangiorgi, Italy (pre-recorded) | Modelling Inherited Retinal Diseases with Stem Cells: Michael Cheetham, UK | Diversity in Genetic Studies aids Locus Discovery, Fine-Mapping, Gene Prioritisation, and Causal Inference: Karoline Kuchenbaecker, Genomics England | GestaltMatcher and DeepGestalt : powerful engines for clinical and molecular diagnostic : Peter M Krawitz, Germany | Single Cell Transcriptomics: Peter Kharchenko, USA (pre-recorded) | Newborn Screening: Way Forward for SDGS From Global Perspective: Carmencita Padilla, Philippines | |
| | Importance of Building up Local Infrastructure to Provide Whole-Genome Sequencing for Rapid Response, for Critically Ill Patients: Ahmad Abou Tayoun, United Arab Emirates | The African Genomic Medicine Training (AGMT) Initiative: Nicola Mulder, South Africa | Therapeutic approaches for genetic conditions: Ethical and social considerations: Nchangwi Syntia Munung, South Africa | Genetic Therapies for Inherited Retinal Diseases: From Fiction to Fact: Bart Leroy, Belgium | Incorporating Local Ancestry in the Construction of Polygenic Risk Scores: Yun Li, USA (pre-recorded) | Objective evaluation of dysmorphisms by automated analysis of facial photographs in African individuals: Aimé Lumaka, DR Congo | Gut microbiome: Alexandra Zhernakova, Netherlands | Next Generation Sequencing Technology: Levelling the Playing Field For SDGS in Genomic Medicine: Lai Poh San, Singapore | |
| | | Educating the Genomics Community on Updated ACMG/Clingen Technical Standards for Interpretation and Reporting of Constitutional Copy Number Variants: Erin Rooney Riggs, USA | | | | | | | |
| | | Panel discussion with all presenters: Lessons Learned from International Workforce Development Initiatives: Looking Back to Move Forward | | | | | | | |
| 12:30 - 14:00 | Lunch Break | | | | | | | | |
| 13:00 - 14:00 | LUNCHTIME SYMPOSIA / WORKSHOPS | Lunchtime Symposium 5 | Lunchtime Symposium 6 | Lunchtime Symposium 7 | Lunchtime Symposium 8 | Africa Population Cohorts Consortium (CE-APCC) (Meeting Block 1.60, Level 1) | Workshop / Focus Group 10 | Workshop / Focus Group 11 | Workshop / Focus Group 12 |
| 14:00 - 15:00 | POSTER SESSION (Venue: Exhibition Hall - Hall 1 & 2, Ground Level) | | | | | | | | |
| 15:00 - 15:30 | Tea / Coffee Break | | | | | | | | |
| th | Session Chairs | MEETING ROOM : Ballroom East | MEETING ROOM : Room 1,61 - 1,62 | MEETING ROOM : Room 1,41 - 1,42 | MEETING ROOM: 1,43 - 1,44 | MEETING ROOM:1,63 - 1,64 | MEETING ROOM: 2,6 | MEETING ROOM: 2,44 - 2,46 | MEETING ROOM: 2,41 - 2,43 |
| 15:30 - 16:00 | Invited Keynote Speakers | Jozef Gez and Maria Mudau Keynote 9 - From Chromosomal Microarray Analysis to a Complete Genome Scan: the ever-evolving lessons from Prenatal Diagnosis: Richard Choy, China | Soudad Rahmouni and Juliana Miranda Cerqueira Keynote 10 - How digital tools are changing equity & quality of genomic medicine: Evidence, preferences & policy: Yvonne Bombard, Canada | Deborah Morris-Rosendahl and Khuthala Mnika Keynote 11 - Consanguinity and genetic diseases in North Africa: are we ready for precision medicine? Cherine Charfeddine, Tunisia | MK Thong and Ileana-Delia Sabau Keynote 12 - Genomics Equity for Health in Latin America: Claudia Gonzaga-Jauregui, Mexico | Melanie Newport and Mridul Johari Keynote 13 - Using Zebrafish Larvae to Characterize Candidate Genes for Cardiometabolic Health: Marcel den Hoed, Sweden | Leon Mutesa and Rhys Dore Keynote 14 -Using Epigenomics to Understand Human Disease: John Greally, USA | Margaret Pericak-Vance and Nadia Carstens Keynote 15 - Establishing an African-relevant roadmap for prostate cancer precision medicine: lessons from whole genome profiling within the Southern African Prostate Cancer Study (SAPCS): Vanessa Hayes, Australia | Pilar Carvalho and Rita Matos Keynote 16 - Rare variants and common traits: Alex Reymond, Switzerland |
| | Oral Presentations | | | | | | | | |
| 16:00 - 16:15 | | OP049. Diagnosing rare genetic disorders in LMICs using WES and automated deep phenotyping and its implications on public health: Alexej Knäus, Germany | OP055. Large-scale single-cell RNA sequencing of the burden of birth defects: Kathleen Strong, Switzerland | OP061. WHO and Partners present a revision of the burden of birth defects: Kathleen Strong, Switzerland | OP067. Maximizing Participation in Genomics Research; Feedback from the Community: Daniel Sekwo, Ghana | OP073. Characterizing 99 cardiometabolic candidate genes for a role in NAFLD using CRISPR/Cas9, in vivo imaging and deep learning in zebrafish larvae: Endrina Mujica, Sweden | OP079. Epigenetic age acceleration in the emerging burden of cardiometabolic diseases among migrant and non-migrant African populations: the population based cross-sectional RODAM study: Felix Chilunga, Netherlands | OP085. Heterogeneous genetic architectures of prostate cancer in African populations: Joseph Lachance, USA | OP091. A multi-omics classifier for prediction of Androgen treatment response in prostate cancer patients: Itunoluwa Isewon, Nigeria |
| 16:15 - 16:30 | | OP050. Diagnostic yield of massive parallel sequencing in postmortem genetic analyses of sudden unexplained deaths in the young Kazakhstani individuals: Ainur Akilzhanova, Kazakhstan | OP056. PheWAS-based clustering of Mendelian Randomization instruments reveals distinct mechanism-specific causal effects between obesity and educational attainment: Liza Darrous, Switzerland | OP062. Breast cancer patients from Inkosi Albert Luthuli Central Hospital in South Africa: comparison of clinical and molecular characteristics between triple-negative and luminal-like disease: Herculaas MvE Combrink OBO Jaco Oosthuizen, South Africa | OP068. A staged approach to developing the genomics workforce while implementing genomic medicine: Amy Nisselle, Australia | OP074. Large-scale analysis of retinal vascular parameters from UK BioBank colour fundus images reveals their phenotypic and genotypic correlations, as well as genes and pathways associated with vascular pathomechanics and diseases: Sven Bergmann, Switzerland | OP080. Genomewide longitudinal DNA methylation profiling of pregnant women and its association with preterm birth outcome – a GARBH-Int study: Jagyashila Das, India | OP086. Rare, risk-associated variants in DNA damage repair genes identified through germline sequencing in two hereditary prostate cancer cohorts: Georgea Foley, Australia | OP092. Application of Machine Learning to predict prostate cancer aggressiveness – using clinical and genomic datasets: Shakuntala Baichoo, Mauritius |
| 16:30 - 16:45 | | OP051. Rapid and comprehensive diagnostic method for repeat expansion diseases using nanopore sequencing: Naomichi Matsumoto, Japan | OP057. Assessment of 3D genomic features across multiple cell types reveals novel insights into the pathogenesis of childhood obesity: Khanh B. Trang, USA | OP063. Genome-wide association study identifies common variants associated with breast cancer in South African black women: Mahtaab Hayat, South Africa | OP069. The Common Infrastructure for National Cohorts in Europe, Canada, and Africa (CINECA) Project: Thomas Keane, UK | OP075. The Genetic Architecture of the Corpus Callosum and its Subregions: Megan Loraine Campbell, South Africa | OP081. Epigenome-wide association study reveals CpG sites associated with thyroid function and regulatory effects on KLF9: Alexander Teumer, Germany | OP087. The role of genetics in colorectal carcinogenesis; Does race matter?: Hassan Ashktorab, USA | OP093. Benchmarking of univariate pleiotropy detection methods: Michael Nothnagel |
| 16:45 - 17:00 | | OP052. Combination of glass-needle-based chromosome microdissection and bisulfite sequencing to assess allele-specific DNA methylation data in unbalanced X-autosome translocations: Bianca Favilla, Brazil | OP058. Development of a pathology-supported genetic testing service at the intersection of research and oncology practice: Translating population risk into personal utility using point-of-care genomics and next generation sequencing: Maritha Kotze, South Africa | OP064. Impact of DNA repair genes on reproduction and cancer predisposition: Svetlana Yatsenko, USA | OP070. The GIRDA project: promoting precision ophthalmology in Africa: Lisa Roberts, South Africa | OP076. The Genetic Architecture of Amygdala Nuclei: Mary Mufford, South Africa | OP082. cis-eQTL mapping of TB-T2D comorbidity elucidates the involvement of African ancestry in TB susceptibility: Yolandi Swart, South Africa | OP088. Genetic ancestry correlates of the somatic mutational landscape from tumor profiling data of 100,000 cancer patients: Francisco De La Vega, USA | OP094. Identifying genes involved in hip osteoarthritis and describing the cells in which they are differentially expressed: Kaitlyn Flynn, Australia |
| 17:00 - 17:15 | | OP053. Stepwise ABC system for classification of any type of genetic variant: Experience so far and suggested updates: Gunnar Douzgos Houge, Norway | OP059. The influence of autozygosity on disease across the phenotypic spectrum: Hilary Martin OBO Daniel Malawsky, UK | OP065. Confined placental mosaicism and clinical impact, a retrospective cohort analysis: Diane Van Opstal, Netherlands | OP071. Development and evaluation of a decision aid for BRCA1/2 genetic testing in high-risk Malaysian families with varying degrees of health literacy: A mixed-methods study: Bettina Meiser, Australia | OP077. Understanding genetic contributions to the brain building blocks of musical rhythm traits: Reyna Gordon, USA | OP083. 3D genome architecture in frontal cortex suggests novel mechanisms for ancestry-specific genetic risk for Alzheimer's Disease: Liyong Wang, USA | OP089. ancMETA: Bayesian Framework for Gene/Subnetwork-Specific Meta-Analysis: Joel Defo, South Africa | OP095. Neurobiological Pathways Underlying Opioid Addiction Identified by Systems Biology Multi-omic Gene Integration: Daniel Jacobson, USA |
| 17:15 - 17:30 | | OP054. Clinical Genomics in Southern Africa: Lessons from the Undiagnosed Disease Programme: Shahida Moosa, South Africa | OP060. Novel genetic associations underlying symptom severity and infection susceptibility to SARS-CoV-2: Mattia Cordiali, Finland | OP066. Novel gene discovery for Parkinson's disease in a South African family: Katelyn Cuttler, South Africa | OP072. The Baylor College of Medicine Genomic Research to Elucidate the Genetics of Rare (BCM-GREGoR) Disease Program: Annotating Gene Function to Provide Insights into Human Biology and Disease Across Worldwide Populations: Jennifer Posey, USA | OP078. Replication of Suggestive GWAS Signals Identified Novel Loci for Non-syndromic Orofacial Clefts in the African Population: Azeez Alade, USA | OP084. Exploring temporal gene expression dynamics during postnatal human brain maturation: a single nucleus RNA sequencing study of the paediatric and adult human brain: Christina Steyn, South Africa | OP090. Using a population-specific reference panel improves genotype imputation accuracy in individuals of African ancestry: Richard Mayanja | OP096. Copy number variant identification from exome sequencing data – a possible approach for African developmental disorder datasets?: Nadja Louw, South Africa |

18:00 - 19:30

HUGO Gathering Event (Meeting Block 1.60)

GenQA Dinner Symposium: How to deliver a high quality genomic diagnostic testing service - reporting results, standardising nomenclature and quality assurance. (Meeting Block 2.60)

Saturday, 25 February 2023

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| 07:30 - 17:00 | Registration open (entrance to Hall 1, Ground Level) | | | | | | | | |
| 09:00 - 16:00 | Exhibition open (Hall 1 & 2, Ground Level) | | | | | | | | |
| 07:30 - 08:45 | Meet the Professor / Meet the Expert / Workshops / Focus Groups / Breakfast Symposia Non Profit Organisations, Academic Institutions and Individuals applications (closing date: 30 November 2022): https://www.ichg2023.com/workshops-satellite-meetings-2/ Sponsor and Exhibitor Applications (apply for a workshop / symposium sponsorship: https://www.ichg2023.com/sponsorship/) | | | | | | | | |
| 09:00 - 10:30 | Plenary 3 - Mechanisms of Disease: The Functional Genome Chairs: Poh San Lai and Inga Prokopenko Venue: Ballroom, Level 1 | | | | | | | | |
| 09:00 - 09:45 | "Autozygome as a Tool to Enhance the Clinical Annotation of the Human Genome" - Fowzan Alkuraya, Saudi Arabia | | | | | | | | |
| 09:45 - 10:30 | Genetics and epigenetics of pediatric epilepsy: moving from etiology to precision therapy - Heather Mefford, USA | | | | | | | | |
| 10:30 - 11:00 | Tea / Coffee Break | | | | | | | | |
| 11:00 - 12:30 | INVITED SESSIONS | Session 17 - Predicting Phenotypes for Polygenic Traits in Diverse Populations Chair: Ricardo Vrdugo, Chile (Venue: Meeting Block 2,41 - 2,43 , Level 2) | Session 18 - The Role of Patient Advocacy - The Example of Rare Disease Chairs: Helen Malherbe, South Africa & Mariela Larrandaburu, Uruguay (Venue: Meeting Block 1,61 -1,62 , Level 1) | Session 19 - Long read genomics Chair: Naomichi Matsumoto, Japan (Venue: Meeting Block 2,6, Level 2) | Session 20 - Inherited Errors of Immune Deficiency: Primary Immune Deficits Chairs: Craig Kinnear, South Africa (Venue: Meeting Block 1,43 - 1,44, Level 1) | Session 21 - Overcoming Barriers to Capturing Diversity in Global Genetics Research Chair: Athena Starlard-Davenport, USA (Venue: Meeting Block 1,63 - 1,64, Level 1) | Session 22 - Shaping the Global Landscape and Developing a Common Language and Standards for Human Genetics and Genomics Chair: Michele Ramsay, South Africa (Venue: Ballroom East, Level 1) | Session 23 - Artificial Intelligence to Enable Clinical Genomics Around the World Chair: Francisco De La Vega, USA & Karen Eliback, USA (Venue: Meeting Block 2,44 - 2,46 Level 2) | Session 24 - Reproductive Carrier Screening - Meeting the Challenges Chair: Edwin Kirk, Australia (Venue: Meeting Block 1,41 - 1,42, Level 2) |
| | | Assessment of the Predictive Power of Polygenic Risk Scores in an Admixed Population from South America for Nine Common Diseases: Ricardo A. Verdugo, Chile | Orphanet and hospital-based programs as tools to address undiagnosed and rare diseases: Francesc Palau, Spain | A catalogue of human structural variations: Shinichi Morishita, Japan | NGS and Infectious Diseases - A Journey of Discoveries: Anne Puel, France (prerecorded) | Diversity in Genetics and Genomic Studies: Adebowale A. Adeyemo, USA | HUGO, The Human Genome Organization, Working to Ensure that the Human Genome Includes and Benefits All: Ada Hamosh, USA | Identifying facial phenotypes of genetic disorders using deep learning: Peter Krawitz, Germany | Ethical aspects of population-scale reproductive genetic carrier screening: Ainsley Newson, Australia |
| | | Developing Effective Polygenic Risk Scores for Cancer for Latin American Populations: Luis Carvajal Carmona, USA | The importance of collaboration between the lay support communities and research groups for rare disease research: Colleen Aldous, South Africa | Long-read genomics to unravel undiagnosed rare diseases: Alexander Hoischen, The Netherlands | WES and WGS in South Africa for Inherited Errors of Immunity – Facts, Fallacies and VUS: Brigitte Glanzmann, South Africa | Beyond Inclusion: Empowering Indigenous Genomic Data Equity in Health: Krystal Tsosie, USA | Developing Technical Standards and Policy Frameworks to Accelerate Responsible Sharing of Genomic and Related Health Data: Peter C Goodhand, Canada | Artificial intelligence for near instant diagnosis of rare genetic disease from whole-genome and deep phenotype data: Martin Reese, USA | Turning the tide on autosomal recessive diseases: The double edged sword of consanguinity: Fowzan Alkuraya, Saudi Arabia |
| | | Developing Effective Polygenic Risk Scores for Multi-Ethnic Populations in Africa: Eimear Kenny, USA | The strength of the associative movement in rare diseases in Ibero-America: ENSERIO LATAM study, future challenges: Juan Carrion, Spain | Strategy detecting abnormal repeats in human diseases: Naomichi Matsumoto, Japan | IEI & Genetics: Clinical illustrations of the Southern African challenges' Andre van Niekerk, South Africa | Building capacity in African Genomics: perspectives from an early career researcher: Lerato Majara, USA | Realizing the Full Potential of Longitudinal Populations Studies and Health Systems to Advance Global Health: Geoffrey S Ginsburg, USA | Histogenomics: Prediction of cancer somatic mutations, tumor mutational burden, and molecular subtypes from histopathological images by deep learning: Francisco De La Vega, USA | Design, implementation and outcomes of The Australian Reproductive Genetic Carrier Screening Project ("Mackenzie's Mission") – screening > 9,000 couples for 1,281 genes to pave the way for a national screening program: Edwin Kirk, Australia |
| | | | The Power of Advocacy: Marianne Gomes, South Africa | | | | | | |
| 12:30 - 14:00 | Lunch Break | | | | | | | | |
| 13:00 - 14:00 | LUNCHTIME SYMPOSIA / WORKSHOP | Lunchtime Symposium 5 | Lunchtime Symposium 6 | Lunchtime Symposium 7 | Lunchtime Symposium 8 | Workshop / Focus Group 9 | Workshop / Focus Group 10 | Workshop / Focus Group 11 | Workshop / Focus Group 12 |
| 14:00 - 15:00 | POSTER SESSION (Venue: Exhibition Hall - Hall 1 & 2, Ground Level) | | | | | | | | |
| 15:00 - 15:30 | Tea / Coffee Break | | | | | | | | |
| 15:30 - 16:00 | Session chairs Zane Lombard and Mohamed Alimohamed | MEETING ROOM: Meeting Room 1,63 - 1,64 Zane Lombard and Mohamed Alimohamed | MEETING ROOM : Room 1,61 - 1,62 Cairna Schlebusch and James Wilson | MEETING ROOM : Room 1,41 - 1,42 Hugues Abriel and Jennifer Posey | MEETING ROOM: 2,6 Colleen Aldous and Elena Avram | MEETING ROOM: Ballroom East Andrew Morris and Yosuke Tanigawa | MEETING ROOM: 2,41 - 2,43 Collet Dandara and Oppah Kuguya | MEETING ROOM: 2,44 - 2,46 Philip Awadalla and Wisdom Akurugu | MEETING ROOM: 1,43 - 1,44 ed |
| 15:30 - 16:00 | Invited Keynote Speakers Keynote 17 - Heterotaxy: the Evo-Devo of Left-Right patterning across vertebrates: Bruno Reversade, Singapore | Keynote 18 - Populations genetics and evolution - A Mexican cohort: Mashaal Sohail, Mexico | Keynote 19 - Genetic bases of severe childhood speech disorders: Angela Morgan, Australia | Keynote 20 - Enabling the ethical return of genomic research results: a global perspective: Danya Vears, Australia | Keynote 21 - Challenges in biobank-based personalized risk prediction and communication: Krista Fischer, Estonia | Keynote 22 - Ethical challenges of cutting-edge technologies: Ursula Matte, Brazil | Keynote 23 - Bioinformatics for Personalized Medicine in Estonian Biobank: Reedik Magi - Estonia | Keynote 24 - Recent progress in the genetics of complex traits with special reference to human twinning: Nick Martin, Australia | |
| 16:00 - 16:15 | Oral Presentations OP097. Deciphering Developmental Disorders in Africa (DDD-Africa): clinical characterization in a Central African setting: Prince Makay, DRC (Congo) | OP103. Expansion of pastoralism across the African continent from a genomic perspective: Cesar Fortes-Lima, Sweden | OP109. Age Estimate of GJB2-p.(Arg143Trp) Founder Variant in Hearing Impairment in Ghana, Suggests Multiple Independent Origins across Populations: Elvis Twumasi Aboagye, Ghana | OP115. Trustworthy health data governance and the social license for precision medicine: Outcomes from mixed methods research in Singapore (2018-2021): Tamra Lysaght, Singapore | OP121. Use of exome sequence data from 200,000 UK Biobank subjects to elucidate the rare variant contribution to common phenotypes: David Curtis, UK | OP127. Building an African-specific pangenome based on short-read whole genome sequence data: Shaun Aron, South Africa | OP133. Transcriptome signature of sodium intake in and links to cardiovascular traits: Amadou Gaye, USA | OP139. Trans-ancestry GWAS meta-analysis of random glucose provides insights into diabetes pathophysiology, complications, and treatment stratification: Marika Kaakinen, UK | |
| 16:15 - 16:30 | OP098. Recurrent variants in subunits of the Human Mediator complex affect brain development and lead to severe neurodegenerative diseases: Elisa Cali, United Kingdom | OP104. Precolonial sex-biased admixture in Indigenous Americans living on the eastern slopes of the Andes: Victor Borda, USA | OP110. Long-read sequencing and profiling of RNA-binding proteins reveals the pathogenic mechanism of aberrant splicing of an SCN1A poison exon in individuals with epilepsy: Hannah Happ, USA | OP116. Democratizing sickle cell disease gene therapy knowledge: A community based model for stakeholder engagement: Vence Bonham, USA | OP122. 300 Billion Associations: Genetic architecture of >2000 phenotypes in 658,000 individuals of diverse ancestries in the VA Million Veteran Program: Anurag Verma, USA | OP128. Individuals of African ancestry at the APOE ε4 gene have a lower risk effect for Alzheimer disease potentially due to gene expression and chromatin accessibility profiles: Katrina Celis, USA | OP134. Non-Mendelian inheritance patterns and extreme deviation rates of CGG short tandem repeats in autism: Dale Annear, Belgium | OP140. Multi-ancestry genome-wide association study in >2.5 million individuals reveals distinct biological pathways driving type 2 diabetes susceptibility with heterogeneous effects across diverse population groups: Eleftheria Zeggini, Germany | |
| 16:30 - 16:45 | OP099. Spliceosome malfunction causes neurodevelopmental disorders with autistic features: Dong Li USA | OP105. Genetic diversity in immune receptor genes in African populations: Yasmina Jaufferally-Fakim, Mauritius | OP111. Genetic, molecular and mouse model investigations of broad neurodevelopmental impact of deleterious variants of the TREX mRNA export complex subunits: Jozef Geck, Australia | OP117. Consanguinity and rare recessive variants involved in multifactorial traits in non-European descent individuals from UK Biobank: Anne-louise Leutenegger, France | OP123. Subcontinental admixture in Europeans and European Americans: Implications for genetic epidemiology studies: Amy Bently OBO Mateus Gouveia, USA | OP129. Characterisation of pharmacogene haplotype variation in African populations: David Twesigomwe, South Africa | OP135. A meta-analysis algorithm called Meta-Analysis by Information Content algorithm is used to prioritise SNPs associated with susceptibility to COVID-19: Bo Wang, UK | OP141. Genetics of metabolic dysfunction through lenses of insulin resistance using GWIS: Inga Prokopenko OBO Liudmila Zudina, UK | |
| 16:45 - 17:00 | OP100. DHX9, the gene encoding the DExH-box helicase DHX9, underlies neurodevelopmental disorders and Charcot-Marie-Tooth disease: Daniel Calame, USA | OP106. Structure and origin of genetic diversity in Tuvans revealed by genome-wide SNPs and Y-chromosomal haplogroups: Vadim Stepanov, Russian Federation | OP112. Selection acting on somatic structural variation in blood impacts molecular function and cancer risk among humans: Kimberly Skead, Canada | OP118. Adaptation and Implementation of Cancer Genetics Testing and Counseling in Africa: Achille Van Christ Manirakiza, Rwanda | OP124. Indigenous American ancestry composition of reference panel causes different imputation performance in two distinct Latin American cohorts: Jennifer French-Kwawu, UK | OP130. Pharmacogenomics of Warfarin: Black Africans Portray Unique Profiles Important for an African-Specific Dosing Algorithm: Arinoo Ndadza, South Africa | OP136. Elucidating a molecular mechanism linking infectious disease exposure and psychosis using multi-omic schizophrenia datasets: Conrad Iyegbe, USA | OP142. Integrating transcriptomics and methylomics with genome wide association analyses reveal novel insights into obesity in Africans: Guanjie Chen, USA | |

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| 17:00 - 17:15 | | OP101. Genomic characteristics of Rett syndrome modifier genes: <i>Alana Slike, Canada</i> | OP107. Genome-wide association study identifies novel candidate malaria resistance genes in Cameroon: <i>Kevin Esoh, South Africa</i> | OP113. Monogenic causes of Premature Ovarian Insufficiency are likely rare and mostly recessive: <i>Saleh Shekari, UK</i> | OP119. Feedback of individual research results in neurodevelopmental disorders in South-Africa: Challenges encountered when bridging the research-diagnostic gap: <i>Zandre Bruwer, South Africa</i> | OP125. The effect of family history and polygenic risk scores on obesity in the Lifelines Cohort Study: <i>Rujia Wang, Netherlands</i> | OP131. GSTM1 and GSTP1 polymorphisms as pharmacogenetic markers for relapse and peripheral neuropathy among African cervical cancer patients on cisplatin treatment: <i>Oppah Kuguyo, Zimbabwe</i> | OP137. Tuberculosis (TB) surveillance in wastewater using the metatranscriptomic approach: <i>Hlengiwe Nombuso Mtetwa, South Africa</i> | OP143. Anthropometric and cardiometabolic traits in Southern African populations are strongly impacted by Khoe-San ancestry and its interaction with lifestyle factors: <i>Dhriti Sengupta, South Africa</i> |
| 17:15 - 17:30 | | OP102. Identification of epigenatures in overgrowth disorders with intellectual disability: A novel approach for genomic diagnostic confirmation: <i>Jair Antonio Tenorio Castaño, Spain</i> | OP108. Characterizing mosaic chromosomal alterations in an African cohort: <i>Jonathan Evans, South Africa</i> | OP114. Combating Obesity by Modulating Lipid Accumulation: In Search of Novel Molecular Regulators of Adipogenesis: <i>Melvin Ambele, South Africa</i> | OP120. Shingles infection and recurrent miscarriage – using genetics to dissect shared genetic susceptibility and causal relationships: <i>Yevheniya Sharhorodska, Ukraine</i> | OP126. African-specific genetic loci determine iron status in African children: <i>John Muriuki, Kenya</i> | OP132. Ex vivo studies of a rare TP53 germline variant (NM_001126114.2, c.1018A>G, p.N340D): Peripheral blood mononuclear cells completely resist cell death induced by the anthracycline, doxorubicin: <i>Claudia Christowitz, South Africa</i> | OP138. The dark side of short-read whole genome sequencing: exploring camouflaged gene regions in African genomes: <i>Melissa Nel, South Africa</i> | OP144. Discovery and fine-mapping of kidney function loci in 80 000 African ancestry individuals: <i>Christopher Kintu, Uganda</i> |
| 18h30 - 22h30 | Conference Dinner at GOLD Restaurant (A true African experience) Ticketed event: R1,150 per person Limited to 350 places | | | | | | | | |

Sunday, 26 February 2023

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| 07:30 - 17:00 | Registration open (entrance to Hall 1, Ground Level) | | | | | | | | | |
| 09:00 - 16:00 | Exhibition open (Hall 1 & 2, Ground Level) | | | | | | | | | |
| 07:30 - 08:45 | | | | | | | | | | |
| 09:00 - 10:30 | Plenary 4 - The future of genetics and genomics Chairs: Ambroise Wonkam and Adebawale Adeyemo (TBC) Venue: Ballroom, Level 1 | | | | | | | | | |
| 09:00 - 09:45 | "Interpreting Genomic Variation in Cancer " - Mathieu Lupien, Canada | | | | | | | | | |
| 09:45 - 10:30 | "The Next Frontier in Personal Genomics" - Eric Topol, USA (pre-recorded) | | | | | | | | | |
| 10:30 - 11:00 | Tea / Coffee Break | | | | | | | | | |
| 11:00 - 12:30 | INVITED SESSIONS | Session 25 - International Genetic Epidemiology Society: Genomics for Global Precision Medicine Chair: Eleftheria Zeggini, Germany (Venue: Meeting Block 2,6 Level 2) | Session 26 - Global Globin Network (GGN): (Public Health Issues in Haemoglobinopathies) Chair: Ghada El-Kamah, Egypt (Venue: Meeting Block 1,61 - 1,62, Level 1) | Session 27 - Resolving Unresolved Neurodevelopmental Disabilities Chairs: Jozef Gecz, Australia & Heather Mefford, USA (Venue: Meeting Block 1.41 - 1,42 Level 1) | Session 28 - Southern African Population Structure, Admixture and Adaptation Chair: Carina Schlebusch, Sweden (Venue: Meeting Block 143 - 1,44 Level 1) | Session 29 - Polygenic Risk Scores In Practice: Laboratory, Clinical, And Ethical Perspectives Chair: Jason Vassy, USA (Venue: Ballroom East, Level 1) | Session 30 -Precision Molecular Cytogenetics: A Time to Sequence Chair: Richard Kwong Wai Choy, China (Venue: Meeting Block 2.41 - 2,43, Level 2) | Session 31: Showcasing H3Africa research Chair: Zane Lombard, South Africa (Venue: Meeting Block 2.44 - 2.46, Level 2) | Session 32 - Privacy, consent and data sharing in the international genetics community Chairs: Francesca Forzano, UK & Kelly Ormond, Switzerland/USA (Venue: Meeting Block 1,63 - 1,64, Level 1) | |
| | | Genomics of Metabolic Disease in Diverse Populations: Inês Barroso, UK | Genetic diversity of thalassaemia in Southeast Asia: Ezalia Esa, Malaysia | Current day clinical genetics in neurodevelopmental disorders: Tjitske Kleefstra, Netherlands. | The Complexity of Population Movements and Admixture Patterns during the Bantu Expansion: Sandra Beleza, UK | Polygenic Risk Scores in the Clinical Laboratory: Matthew Lebo, USA (pre-recorded) | Mate-Pair Sequencing and the Road towards Comprehensive Identification and Interpretation of Pathogenic Structural Variation: Cynthia C Morton, USA | Intergenerational and epigenetic effects of trauma and PTSD following exposure to the 1994 genocide against the Tutsi in Rwanda: Leon Mutesa, Rwanda | Genomic data sharing in Africa with perspectives from H3Africa and the South African Protection of Personal Information Act: Michele Ramsay, South Africa | |
| | | Polygenic risk scores across diverse populations: Alisa Manning, USA | Modern therapeutics approaches in Beta Thalassaemia: Ghada El-Kamah, Egypt | DNA Methylation Episignatures as Clinical Diagnostic Biomarkers for Rare Diseases: Bekim Sadikovic, Canada | Khoisan Gene Flow in Southern Africa and Implications for Genetic Studies: Ananyo Choudhury, South Africa | Polygenic Risk Scores in Medical Practice: Jason Vassy, USA | Implementation Of Genome Sequencing: Experience in a Genetic Diagnostic Laboratory: Zirui (Elvis) Dong, China (pre recorded) | Neurogenetics study in Mali: new discoveries and perspectives: Guida Landoure, Mali | The Brazilian Initiative on Precision Medicine (BIPMed): experiences on sharing genomic information in Brazil and Latin America for the past seven years: Iscia Lopes-Cendes, Brazil | |
| | | Using Genomics for the Treatment and Prevention of Breast Cancer: Olufunmilayo Olopade, USA | Human Genetics and Health: Gene Therapy for Sickle Cell Disease in Africa: Julie Makani, Tanzania (pre-recorded) | Rare and Common Variant Contributions to NDD: Hilary Martin, UK | The Evolution of Skin Pigmentation in Southern Africa: Dana Al-Hindi, USA | Ethical Considerations of Polygenic Risk Scores in the Clinic: Anna C.F. Lewis, USA | Diagnostic Management of Apparently Balanced Structural Variants: Matthew Chau, USA | Ten years of AWI-Gen: insights from genomic studies and the way forward: Dhriti Sengupta, South Africa | Research and Policy initiatives in Australia regarding genomic data sharing: Ainsley Newson, Australia | |
| | | | | | | | | | Round Table discussion | |
| 12:30 - 13:30 | Closing Ceremony Chairs: Chrales Rotimi, Raj Ramesar and Michele Ramsay (Venue: Ballroom, Level 1) - Awards - Best Oral - Awards - Best Poster - Awards - Lifetime Achievement - Handover to next host country and the next President of the IFHGS | | | | | | | | | |