Thursday, 23 February 2023

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 / Meet the Expert / Workshops / Focus Groups / Breakfast Symposia**								
09:00 - 10:30	**Plenary 1 - Genomics and anthropology - insights on the peopling of Africa** Venue: Ballroom (Level 1) Chairs: Michele Ramsay and Charles Rotimi								
09:00 - 09:45	"The genetic history of Africa based on modern and ancient DNA " - Carina Schlebusch, Sweden								
09:00 - 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: Ballroom West, Level 1)	**Session 3 - Hemoglobinopathies: Challenges and Perspectives** Chair: Martine de Rycke, Belgium (Venue: Meeting Block 1.4, Level 1)	**Session 4 - Vascular Anomalies: from Somatic Mutations towards Targeted Therapies** Chair: Miikka Vikkula, Belgium (Venue: Meeting Block 1.6, 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 2.41-2.43, Level 2)	**Session 6 -The future of Genomics (ClinGen and ClinVar)** Chair: Erin Rooney Riggs, USA (Venue: Meeting Block 2.44 - 2.46, Level 2)	**Session 7 - Cancer Genetics** Chair: Raj Ramesar, South Africa (Venue: Meeting Block 2.61-2.63, Level 2)	**Session 8 - Susceptibility to Infectious Disease - from genetics to exposome** Chairs: Inga Prokopenko, UK & Brent Richards, Canada (Venue: Meeting Block 2.64 - 2.66, 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	ClinVar: Perspectives from Global Users/Submitters: João Bosco de Oliveira Filho, Brazil	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	Lessons learned in implementing the neuropsychiatric genetics capacity building program, GINGER: Kristianna Post, USA	The Clinical Genome Resource (ClinGen): An Overview: Erin Rooney Riggs, USA	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	Utilizing ClinGen Resources: Danielle Azzariti, USA	Cancer Genomics as a measure of Genetic Services in a Developing Country: Raj Ramesar, South Africa	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				Panel: Nadia Carstens, South Africa; Melissa Nel, South Africa; Joao Bosco de Oliveira Filho, Brazil		
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)**	**Lunchtime Symposium 4**	**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**								
	Session Chairs								
15:30 - 16:00	**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 arrythmia 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*

16:45 - 17:00		**OP004**. Deciphering Developmental Disorders in Africa (DDD-Africa) – Clinical lessons learnt following whole exome sequencing in an African setting: *Amanda Krause, South Africa*	**OP010**. Variant-to-gene mapping paired with cross-species validation reveals novel effector genes for insomnia: *Amber Zimmerman, USA*	**OP016**. Non-additive polygenic score: *Rikifumi Ota, Japan*	**OP022.** Novel biological insights into the genetic architecture of bipolar disorder form multi-ancestry genome-wide association studies: *Kevin O'Connell*	**OP028**. Novel Copy Number Variants associated to venous thromboembolism (VTE) and warfarin dose requirement in African Americans: *Carolina Clark OBO Minoli Perera, USA*	**OP034**. Engaging with Local Communities in Botswana: Understanding impact of Cultural Values, Norms and Beliefs on Gene-Editing Research: *Setlhomo Koloi-Keaikitse*	**OP040.** Pleiotropic association of rare variation with the plasma lipidome and type 2 diabetes: *Joanne E Curran, United States*	**OP046**. Genome-wide association study of oesophageal cancer identifies trans-ethnic risk variants in African and Chinese populations: *Wenlong Chen, South Africa*
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: *Maria Mudau, South Africa*	**OP011**. The incorporation of novel audiogram classification strategies to identify genes and pathways involved in age-related hearing loss: *Samah Ahmed, Canada*	**OP017**. BridgePRS: A powerful trans-ancestry Polygenic Risk Score method: *Paul O'Reilly, USA*	**OP023**. Investigating the role of polygenic background in neurodevelopmental disorders: *Qinqin Huang*	**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: *Natàlia Pujol Gualdo, Estonia*	**OP035**. Personal values versus professional role: healthcare professionals experiences with offering termination of pregnancy for fetal abnormalities: *Malebo Malope, South Africa*	**OP041**. Genetic relationships and causality between overall and central adiposity and breast, prostate, lung and colorectal cancer: *Jared Maina, France*	**OP047**. Evidence of potential natural selection in African American individuals post admixture: *James Jaworski, USA*
17:15 - 17:30		**OP006**. Loss-of-function of AMFR causes autosomal recessive hereditary spastic paraplegia by altering lipid metabolism: *Stefan Barakat, Netherlands*	**OP012**. Genome-wide association study of breast density among women of African Ancestry: *Shefali Verma, USA*	**OP018**. Recessive effects of protein-coding variants in a cohort of 44,000 British Pakistanis and Bangladeshis: *Teng Hiang Heng, UK*	**OP024**. The Genetics of Extreme Birthweight and its Relationship with Cardiometabolic Disease: *Gunn-Helen Moen, Australia*	**OP030**. Genomic spectrum of non-syndromic hearing impairment in Senegal: *Rokhaya Ndiaye, Senegal*	**OP036**. Genetic risk prediction of high-risk human papillomavirus infection and cervical cancer: *Triin Laisk, Estonia*	**OP042**. An assessment of the copy number variation landscape in Sub-Saharan African populations: *Laura Cottino, South Africa*	**OP048.** TBD
18:00 - 19:30	Southern African Society of Human Genetics (SASHG) - BGM (Meeting Block 1.41 & 1.42) Workshop: It's more than just training: Capacity building in context. (Hosted by Wellcome Connecting Science) (Meeting Block 1.43 & 1.44)								
19:30 - late	**President's Dinner (by invitation only)**								