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: Charles Rotimi and Raj Ramesar (TBC)								
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 - Host Genomics / Infectious Diseases** Chair: (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 (virtual)	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	Bristol Public health - Nick Timson
		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	HIV transmission - Katherine Atkins (U of Edinburgh)
		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**	**Lunchtime Symposium 1**	**Lunchtime Symposium 2**	**Rara Diseases Celebration day**	**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: Ada Hamosh, USA	Keynote 2 - The genetics of major depression in diverse populations: Karoline Kuchenbaecker, UK	Keynote 3 - Statistical Genetcis & Genetic Epidemiology (complex traits) (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		1. The utility of clinical exome sequencing as a first-tier diagnostic tool in critically ill infants in South Africa: Lisa Campbell, South Africa	1. 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	1. Multi-omics study highlights several metabolites and gene expression levels as predictors of weight loss maintenance: Tingyu Guo, UK	1. 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	1. Detailing the Inflammatory Cascade of Severe Malaria through Transcriptomic, Proteomic, and Metabolomic Analyses: Rafal Sobota, USA	1. 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	1. Development of a polygenic score to predict cisplatin-induced ototoxicity: Deanne Nixie Miao, Canada	1. Core Investigation of Exonic Variations in Olfactory Receptor Genes (ORGs) in Indigenous Populations from Southeast Asia: Sze Mei Lee. Malaysia
16:15 - 16:30		2. Exome Sequencing in Childhood-Onset Essential Hypertension Implicates Monogenic Disruption of Vascular Integrity in Disease Pathogenesis: Edmond Wonkam Tingang, USA	2. 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	2. Circulating cytokines and their role in type 2 diabetes among Africans: A Mendelian randomization study: Karlijn Meeks, USA	2. Targeted delivery of miR-218 via decorated hyperbranched polyamidoamine for liver cancer regression: Mahmoud Elhefnawi, Egypt	2. Investigating the impacts of age at menarche on cognitive function based on a Mendelian randomization approach using a nationwide cohort from the Taiwan Biobank:	2. What are patient perspectives on privacy and trust in digital genomic tools? A qualitative study: Vedika Jha, Canada	2. Large cardiac arrythmia gene panel genetic testing as prognostic tool for Brugada syndrome: Sonia Van Dooren, Belgium	2. 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		3. Investigating Variants Associated with Quantitative Red Blood Cell Phenotypes from TOPMed Predictions in African Sickle Cell Disease Populations: Nabeelah Samie, South Africa	3. Characterizing the shared genetic influences between schizophrenia and reaction time variability: Olivia Wootton, South Africa	3. 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	3. Mismatch Repair variant classification with ACMG/AMP criteria and comparison with Bayesian probability framework: Finlay Macrae, Australia	3. 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	3. Navigating Layers of Uncertainty: Perspectives from South African Genetic Counselling Interactions: Meagan Scott, South Africa	3. Developing an integrated risk score for cardiovascular diseases in African populations: Michelle Kamp, South Africa and UK	3. Global Proteomic Analyses of Type 2 Diabetes and Obesity in West Africans: Adebowale Adeyemo, USA
16:45 - 17:00		4. Designing and evaluating the utility of a panel of de novo mutation enriched genes for diagnosing South African patients with developmental delay: Patracia Nevondwe, South Africa	4. Variant-to-gene mapping paired with cross-species validation reveals novel effector genes for insomnia: Amber Zimmerman, USA	4. Non-additive polygenic score: Rikifumi Ota, Japan	4. Comparing the analytical performance of exome sequencing and traditional panel testing in a cancer population: Emma Reble, Canada	4. Novel Copy Number Variants associated to venous thromboembolism (VTE) and warfarin dose requirement in African Americans: Minoli Perera, USA	4. The Decision-making of at-risk marriages after pre-marital screening for B-thalassemia and Sickle Cell Disease in Al-Qatif, Saudi Arabia: Sara Alkaf, Saudi Arabia	TBD	4. 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		5. 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	5. The incorporation of novel audiogram classification strategies to identify genes and pathways involved in age-related hearing loss: Samah Ahmed, Canada	5. BridgePRS: A powerful trans-ancestry Polygenic Risk Score method: Paul O'Reilly, USA	5. Whole-genome doubling-aware Copy-number Phylogenies for Cancer Evolution: Roland Schwarz, Germany	5. 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	5. Personal values versus professional role: healthcare professionals experiences with offering termination of pregnancy for fetal abnormalities: Malebo Malope, South Africa	5. Genetic relationships and causality between overall and central adiposity and breast, prostate, lung and colorectal cancer: Jared Maina, France	5. Evidence of potential natural selection in African American individuals post admixture: James Jaworski, USA
17:15 - 17:30		6. Loss-of-function of AMFR causes autosomal recessive hereditary spastic paraplegia by altering lipid metabolism: Stefan Barakat, Netherlands	6. Genome-wide association study of breast density among women of African Ancestry: Shefali Verma, USA	6. Recessive effects of protein-coding variants in a cohort of 44,000 British Pakistanis and Bangladeshis: Teng Hiang Heng, UK	6. The Genetics of Extreme Birthweight and its Relationship with Cardiometabolic Disease: Gunn-Helen Moen, Australia	6. Genomic spectrum of non-syndromic hearing impairment in Senegal: Rokhaya Ndiaye, Senegal	6. Genetic risk prediction of high-risk human papillomavirus infection and cervical cancer: Triin Laisk, Estonia	6. An assessment of the copy number variation landscape in Sub-Saharan African populations: Laura Cottino, South Africa	6. Looking for genes influencing non-identical twinning in Africa, Asia and Europe: Nick Martin, Australia
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18:00 - 19:30	Southern African Socieyt of Human Genetics (SASHG) - BGM Non Profit Organisations, Academic Institutions and Individuals applications (closing date: 30 November 2022): https://www.ichg2023.com/workshops-satellite-meetings-2/)								
19:30 - late	**President's Dinner (by invitation only)**								