Saturday, 25 February 2023

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 (TBC) 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	Title TBD - 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: Ballroom East, Level 1)	**Session 18 - The Role of Patient Advocacy - The Example of Rare Disease** Chairs: Helen Malherbe, South Africa & Mariela Larrandaburu, Uruguay (Venue: Ballroom West, Level 1)	**Session 19 - Long read genomics** Chair: Naomichi Matsumoto, Japan (Venue: Meeting Block 1.4, Level 1)	**Session 20 - Inherited Errors of Immune Deficiency: Primary Immune Deficits** Chairs: Craig Kinnear, South Africa (Venue: Meeting Block 1.6, Level 1)	**Session 21 - Overcoming Barriers to Capturing Diversity in Global Genetics Research** Chair: Athena Starlard-Davenport, USA (Venue: Meeting Block 2.61 - 2.63, Level 2)	Session 22 - An Opportunity to Shape the Global Landscape and Develop a Common Language and Standards for Human Genetics and Genomics Chair: Michele Ramsay, South Africa (Venue: Meeting Block 2.44 - 2.46, Level 2)	**Session 23 - Artificial Intelligence to Enable Clinical Genomics Around the World** Chair: Francisco De La Vega, USA & Karen Eliback, USA (Venue: Meeting Block 2.61 - 2.63, Level 2)	**Session 24 - Reproductive Carrier Screening - Meeting the Challenges** Chair: Edwin Kirk, Australia (Venue: Meeting Block 2.64 - 2.66, 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	Long-read genomics to unravel undiagnosed rare diseases: Alexander Hoischen, The Netherlands	NGS and Infectious Diseases - A Journey of Discoveries: Anna Puel, France	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	A catalogue of human structural variations: Shiichi Morishita, Japan	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 solving human repeat diseases: Naomichi Matsumoto, Japan	TBD	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: Kelly du Plessis, South Africa						
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**	**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**								
	Session chairs								
15:30 - 16:00	**Invited Keynote Speakers**	Keynote 17 - Hetrotaxy: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 & computational Approaches (Genome structure) Title TBD: Reedik Magi - Estonia	Keynote 24 - Complex traits & Polygenic Disorders TBD
	Oral Presentations								
16:00 - 16:15		1. Deciphering Developmental Disorders in Africa (DDD-Africa): clinical characterization in a Central African setting: Prince Makay, DRC (Congo)	1. Expansion of pastoralism across the African continent from a genomic perspective: Cesar Fortes-Lima, Sweden	1. Age Estimate of GJB2-p.(Arg143Trp) Founder Variant in Hearing Impairment in Ghana, Suggests Multiple Independent Origins across Populations: Elvis Twumasi Aboagye, Ghana	1. Trustworthy health data governance and the social license for precision medicine: Outcomes from mixed methods research in Singapore (2018-2021): Tamra Lysaght, Singapore	1. Use of exome sequence data from 200,000 UK Biobank subjects to elucidate the rare variant contribution to common phenotypes: David Curtis, UK	1. Building an African-specific pangenome based on short-read whole genome sequence data: Shaun Aron, South Africa	1. Transcriptome signature of sodium intake in and links to cardiovascular traits: Amadou Gaye, USA	1. Trans-ancestry GWAS meta-analysis of random glucose provides insights into diabetes pathophysiology, complications, and treatment stratification: Marika Kaakinen, UK
16:15 - 16:30		2. Deciphering Developmental Disorders in Africa (DDD-Africa) – Clinical lessons learnt following whole exome sequencing in an African setting: Amanda Krause, South Africa	2. Precolonial sex-biased admixture in Indigenous Americans living on the eastern slopes of the Andes: Victor Borda, USA	2. 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	2. Democratizing sickle cell disease gene therapy knowledge: A community based model for stakeholder engagement: Vence Bonham, USA	2. 300 Billion Associations: Genetic architecture of >2000 phenotypes in 658,000 individuals of diverse ancestries in the VA Million Veteran Program: Anurag Verma, USA	2. 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	2. Non-Mendelian inheritance patterns and extreme deviation rates of CGG short tandem repeats in autism: Dale Annear, Belgium	2. 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		3. Spliceosome malfunction causes neurodevelopmental disorders with autistic features: Dong Li USA	3. Genetic diversity in immune receptor genes in African populations: Yasmina Jaufeerally-Fakim, Mauritius	3. Genetic, molecular and mouse model investigations of broad neurodevelopmental impact of deleterious variants of the TREX mRNA export complex subunits: Jozef Gecz, Australua	3. Consanguinity and rare recessive variants involved in multifactorial traits in non-European descent individuals from UK Biobank: Anne-louise Leutenegger, France	3. Subcontinental admixture in Europeans and European Americans: Implications for genetic epidemiology studies: Mateus Gouveia, USA	3. Characterisation of pharmacogene haplotype variation in African populations: David Twesigomwe, South Africa	3. 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	3. Genetics of metabolic dysfunction through lenses of insulin resistance using GWIS: Liudmila Zudina, UK
16:45 - 17:00		4. DHX9, the gene encoding the DExH-box helicase DHX9, underlies neurodevelopmental disorders and Charcot-Marie-Tooth disease: Daniel Calame, USA	4. Genetic Polymorphism of 24 Autosomal STR in the Population of Rwanda:Gasana Paul, Rwanda	4. Selection acting on somatic structural variation in blood impacts molecular function and cancer risk among humans: Kimberly Skead, Canada	4. Adaptation and Implementation of Cancer Genetics Testing and Counseling in Africa: Achille Van Christ Manirakiza, Rwanda	4. Indigenous American ancestry composition of reference panel causes different imputation performance in two distinct Latin American cohorts: Jennifer French-Kwawu, UK	4. Pharmacogenomics of Warfarin: Black Africans Portray Unique Profiles Important for an African-Specific Dosing Algorithm: Arinao Ndadza, South Africa	4. Elucidating a molecular mechanism linking infectious disease exposure and psychosis using multi-omic schizophrenia datasets: Conrad Iyegbe, USA	4. Integrating transcriptomics and methylomics with genome wide association analyses reveal novel insights into obesity in Africans: Guanjie Chen, USA

17:00 - 17:15		5. Genomic characteristics of Rett syndrome modifier genes: Alana Slike, Canada	5. Genome-wide association study identifies novel candidate malaria resistance genes in Cameroon: Kevin Esoh, South Africa	5. Monogenic causes of Premature Ovarian Insufficiency are likely rare and mostly recessive: Saleh Shekari, UK	5. Feedback of individual research results in neurodevelopmental disorders in South-Africa: Challenges encountered when bridging the research-diagnostic gap: Zandre Bruwer, South Africa	5. The effect of family history and polygenic risk scores on obesity in the Lifelines Cohort Study: Rujia Wang, Netherlands	5. GSTM1 and GSTP1 polymorphisms as pharmacogenetic markers for relapse and peripheral neuropathy among African cervical cancer patients on cisplatin treatment: Oppah Kuguyo, Zimbabwe	5. Tuberculosis (TB) surveillance in wastewater using the metatranscriptomic approach: Hlengiwe Nombuso Mtetwa, South Africa	5. Anthropometric and cardiometabolic traits in Southern African populations are strongly impacted by Khoe-San ancestry and its interaction with lifestyle factors: Dhriti Sengupta, South Africa
17:15 - 17:30		6. Identification of episignatures in overgrowth disorders with intellectual disability: A novel approach for genomic diagnostic confirmation: Jair Antonio Tenorio Castaño, Spain	6. Characterizing mosaic chromosomal alterations in an African cohort: Jonathan Evans, South Africa	6. Combating Obesity by Modulating Lipid Accumulation: In Search of Novel Molecular Regulators of Adipogenesis: Melvin Ambele, South Africa	6. Shingles infection and recurrent miscarriage – using genetics to dissect shared genetic susceptibility and causal relationships: Yevheniya Sharhorodska, Ukraine	6. African-specific genetic loci determine iron status in African children: John Muriuki, Kenya	6. 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: Claudia Christowitz, South Africa	6. The dark side of short-read whole genome sequencing: exploring camouflaged gene regions in African genomes: Melissa Nel, South Africa	6. Discovery and fine-mapping of kidney function loci in 80 000 African ancestry individuals: Christopher Kintu, Uganda
18:00 - 19:30	**Meet the Professor / Meet the Expert / Workshops / Focus Groups / Dinner Symposia**								
18h30 - 22h30	**Conference Dinner at GOLD Restaurant (A true African experience)** Ticketed event: R1,150 per person Limited to 350 places								