



PROGRAMME

SATURDAY 3 AUGUST 2019

Young Researchers Symposium (YRS)	08:00 – 16:00		Intaka Island Eco-Centre
Satellite Workshop: Clinical use of genomic tests	09:00 – 16:30	Registration starts at 08.30	Lagoon Beach Hotel Conference Centre, 1 Lagoon Gate Drive, Milnerton
16:00 – 18:00	Main SASHG Conference Registration Venue: Century City Conference Centre		
18:00 – 20:30	Welcome Function Venue: Hall D, Century City Conference Centre		
	Guest speaker: Hendri Herbst The impact of genes and the environment on his life as an S11 paralympic athlete		

SUNDAY 4 AUGUST 2019

Hall D

08:00 – 09:00	Registration	Century City Conference Centre foyer
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OFFICIAL OPENING

09:00 - 09:10	Welcome	Craig Kinnear, LOC Chair
09:10 – 09:20	Welcome	Soraya Bardien, SASHG Chair

SESSION 1: Our genes in the clinical testing environment

Hall C

09:20 – 10:00	Keynote address: Developments in genomics: implications for education and practice	Robert Nussbaum
10:00 – 10:15	Analysis of pathogenic variants for cancer-associated genes in selected African populations	Yasmina Jaufeerally-Fakim
10:15 – 10:30	Development of a cost-effective multi-disease targeted NGS panel for facial dysostoses, Rasopathies and Cohesinopathies (FRASC)	Maria Mudau

10:30 – 11:00

COFFEE BREAK

Hall D

11:00 – 11:30	Keynote address: Use of RNA splicing analysis in genetic/genomic diagnostics	Andrew Douglas
11:30 – 11:45	Genomic analysis of Black South African women with breast cancer	Amanda Krause
11:45 – 12:00	Investigating the allele sequence diversity at the Huntington disease loci in an African population	Jessica Levesley
12:00 – 12:15	The impact of the c.5603A>T hypomorphic variant on ABCA4 founder mutation screening for Stargardt disease in South Africa	Nicole Midgley
12:15 – 12:30	Investigating the genetic aetiology of three facial dysostoses in South Africa	Patracia Nevondwe
12:30 – 12:45	Hereditary colorectal cancer: genetic counselling and testing at Tygerberg Hospital	Mardelle Schoeman
12:45 – 12:50	Rapid Fire Presentation: Next generation sequencing findings in five South African patients with enzymatically confirmed pyruvate dehydrogenase complex deficiency	Surita Meldau
12:50 – 12:55	Rapid Fire Presentation: RRM2B: a candidate gene for Rod-Cone Dystrophy with Sensorineural Hearing Loss and Renal Dysfunction	Lisa Roberts
12:55 – 13:00	Rapid Fire Presentation: Pioneering informed consent in breast cancer genomics towards the return of research results in Africa: The Kenyan experience	Rispah Torroyrey-Sawe

13:00 – 14:00

LUNCH

Hall D

SESSION 2: Our genes in the care and counselling environment

Hall C

14:00 – 14:30	Keynote address: Psychiatric genetic counselling	Jehannine Austin
14:30 – 14:45	Decision-making regarding prenatal testing for Down syndrome and termination of pregnancy: A South African perspective	Chantelle Scott
14:45 – 15:00	Cerebral palsy medico-legal matters in South Africa - A medical geneticist's experience	Arnold Christianson
15:00 – 15:15	Mabry syndrome: using artificial intelligence to diagnose rare genetic conditions	Heidre Bezuidenhout
15:15 – 15:30	Perspectives and experiences of individuals undergoing predictive testing for hereditary breast and ovarian cancer (HBOC) syndrome in the Western Cape, South Africa.	Monica Araujo

15:30 – 16:00

COFFEE BREAK/ POSTER SESSION 1

Hall D

16:00 -16:30	Keynote address: Genetics of Parkinson's disease	Robert Nussbaum
16:30 – 16:45	Lethal multiple pterygium syndrome - a South African case series	Liani Smit
16:45 – 17:00	The reliability and validity of last menstrual period for gestational age estimation in a low-to-middle- income setting	Shelley Macaulay
17:00 – 17:05	Rapid Fire Presentation: Comparison of two testing strategies in hereditary breast and ovarian cancer at Tygerberg hospital	Clair Engelbrecht
17:05 – 17:10	Rapid Fire Presentation: Endocrine profiling in Black South African Fanconi Anaemia patients, homozygous for a FANCG founder mutation	Candice Feben
17:10 – 17:15	Rapid Fire Presentation: The role of a genetic counsellor in a support group for Huntington's disease	Maxine du Toit
17:15 – 17:20	Rapid Fire Presentation: 'It was PGD or no kids!' A qualitative analysis of the patient experience of diagnosis	Bianca Carzis

FREE EVENING FOR SIGHTSEEING/ NETWORKING/ GROUP TEAMBUILDING

MONDAY 5 AUGUST 2019

Psychiatric genetic counselling workshop	07:30 - 16:00	Intaka Island Eco-centre
08:00 – 09:00	Registration	
SESSION 3: Our genes and past environments		Hall C
09:00 - 09:30	Keynote address: The importance of genetic diversity in studies of complex traits	Chris Gignoux
09:30 – 09:45	Sex-specific patterns of inbreeding depression reveal directional dominant genetic architecture of selected complex traits in sub-Saharan populations: an AWI-Gen sub-study	Francisco Ceballos
09:45 – 10:00	Genomic data from >5000 South Africans reveal novel sub-structure reflecting the complex demographic history within South-Eastern Bantu speakers	Dhriti Sengupta
10:00 – 10:15	Tracking migration, admixture and selection patterns in the Indian Ocean trade network	Cesar Fortes-Lima
10:15 – 10:30	Y-chromosome forensics in South Africa: development of a Y-STR profiling system	Maria Eugenia D'Amato
10:30 – 11:00	COFFEE BREAK / POSTER SESSION 2	Hall D
SESSION 4: Our genes in the data environment		Hall C
11:00 – 11:30	Keynote address: The epigenetics of longevity: sirt3, sirt6, sirwanin, you're out	Hugh Patterton
11:30 – 11:45	Transcriptional profiling of antiviral responses at single-cell resolution	Nikki Gentle
11:45 – 12:00	Validation of next generation sequencing for the detection of all pathogenic variants in the familial breast cancer genes BRCA1 and BRCA2	Jaco Oosthuizen
12:00 – 12:15	Genetic determinants of childhood trauma exposure	Shareefa Dalvie
12:15 – 12:30	New insights from a pan-African genome-wide association study with lipid levels: an AWI-Gen sub-study	Ananyo Choudhury
12:30 – 13:30	LUNCH	Hall D
SESSION 5: Our genes in the data environment contd.		Hall C
13:30 – 14:00	Keynote address: The ethical landscape for genomics in Africa	Nicki Tiffin
14:00 – 14:15	Gene-environment interactions with childhood trauma could mediate the relationship between neuroanatomical variation and antipsychotic treatment response	Emma Frickel
14:15 – 14:30	Alcohol's effect on the gut microbiome of pregnant women	Natasha Kitchin
14:30 – 14:45	Development of a custom genotyping platform for detecting prostate cancer associations in men of African descent.	Lindsay Petersen
14:45 – 15:00	Bioinformatic pipelines for transcriptome analyses: Understanding gene expression in black South Africans with systemic sclerosis	Phelelani Mpangase
15:00 – 15:05	Rapid Fire Presentation: Development of a custom-designed targeted resequencing gene panel for Parkinson's disease	Amokelani Mahungu
15:05 – 15:10	Rapid Fire Presentation: Differentially expressed serum microRNAs in South African black type 2 diabetic patients with diabetic nephropathy	Stefan Valentin
15:10 – 15:45	COFFEE BREAK / POSTER SESSION 3	Hall D

15:45 – 16:00	SASHG 2019 Delegates Group photo	Meet in CCC foyer
16:00 – 17:30	SASHG BGM	Meet in Hall C
19:00	SASHG 2019 GALA DINNER: CRYSTAL TOWERS HOTEL	

TUESDAY 6 AUGUST 2019

08:00 – 09:00	Registration	
SESSION 6: Our genes in the community and family environment		Hall C
09:00 – 09:25	Keynote address: The prenatal environment: exposure to teratogens in a Cape Town cohort	Hein Odendaal
09:25 – 09:40	Does smoking modify the effect of alcohol on cardinal facial features? A cohort study	Michael Urban
09:40 – 09:55	An Online Family Health History Tool: System Description and Initial Utilisation	Gareth Kantor
09:55 – 10:10	Modelled data for Oral Facial Clefts in South Africa, 2012	Helen Malherbe
10:10 – 10:30	Rare Diseases South Africa: Links between support groups, researchers and health providers	Kelly du Plessis
10:30 – 11:00	COFFEE BREAK	
SESSION 7		Hall C
11:00 – 12:45	Panel Discussion: Getting Beyond Race Chair: Keymanthri Moodley (Director of the Centre for Medical Ethics & Law)	
12:45 – 13:00	Prize-giving session 1. orals 2. posters 3. rapid-fire talks	
13:00 – 13:15	Wrap-up and closing	Eileen Hoal
13:15 – 14:15	LUNCH and DEPART	