



Global Genomic  
Medicine Collaborative

## Genomic Medicine implementation in low-resource settings

**Young Investigators  
Forum - Agenda**

**28-30  
November  
2018**

President hotel  
Cape Town  
South Africa





## Global Genomic Medicine Collaborative

### **4th International G2MC Meeting** ***Genomic Medicine Implementation in Low Resources Settings*** **November 28-30, 2018**

**President Hotel**  
**President Ballroom**  
4 Alexander Rd, Bantry Bay, Cape Town, 8001, South Africa

#### **Young Investigators' Forum**

#### **DAY 1: NOVEMBER 28, 2018**

08:00-08:05

**Welcoming Remarks - Chair, YIF Planning Committee**  
**Teri Manolio**  
National Human Genome Research Institute, USA

08:05-10:55

#### **Young Investigator Presentations**

*Targeted Next-Generation Sequencing Identifies Novel Variant*  
**Oluwale Oluwafemi**  
Stellenbosch University, Cape Town, South Africa

*African-specific NPHS2 V260E mutation in SR-FSGS cases*  
**Melanie Ann Govender**  
University of Witwatersrand, Johannesburg, South Africa

*Distinct Profiles of warfarin Pharmacogenes in Africans*  
**Arinao Ndadza**  
University of Cape Town, Cape Town, South Africa

*Exploring the Molecular Basis of Hereditary Spinocerebellar Degeneration in a Large Sudanese Family*  
**Arwa Babai**  
University of Kartoum, Sudan

*Novel Alleles of Immunoglobulin Variable Domain in Africa and Broadly Neutralizing Effect against HIV Infection*

**Rahaman Ahmed Ademolu**

University of Lagos, Nigeria

*Using DM Techniques to Predict the Response of HCV Patients*

**Mohammed Ahmed Farahat**

Helwan University, Egypt

*Genome-wide Association Studies, Imputation and Fine Mapping in African Populations Identify Novel Risk Loci for Orofacial Clefts*

**Lord Jephthah Joojo Gowans**

Kwame Nkrumah University of Science and Technology, Ghana

*Genome-wide discovery of long noncoding RNAs in HIV-1 non-pr*

**Stanford Kwenda**

National Inst for Communicable Diseases, Johannesburg, South Africa

10:55-11:10

**COFFEE BREAK**

11:10-11:40

**Career Development Panel**

**Moderator:** Teri Manolio, NHGRI, USA

**Panel members**

**Victoria Nembaware**

University of Cape Town, South Africa

**Gabriela Repetto**

Universidad del Desarrollo, Chile

**Surakameth Mahasirimongkol**

Ministry of Public Health, Thailand

**Cedrik Ngongang**

University of Cape Town, South Africa

11:40-12:00

**Poster Number**

**01**

**Young Investigator Flash Talks**

*GJB2 and GJB6 mutations in non-syndromic childhood hearing impairment in Ghana*

**Samuel Mawuli Adadey**

University of Cape Town and University of Ghana, South Africa

**02**

*PG and PK of CNS penetration of tenofovir/emtricitabine*

**Phumla Sinxadi**

University of Cape Town, Cape Town, South Africa

- 03** *Eleganpro: A Novel Bioinformatics Tool for C. elegans Transcriptome*  
**Olaitan Awe**  
University of Ibadan, Nigeria
- 04** *Profound biotinidase deficiency caused by (D444H) resulting in recurrent early childhood death in a Sudanese Family*  
**Reem Salaheldin Khalid Hamad**  
University of Khartoum, Sudan
- 05** *Microsatellite Instability:NR21, NR24 in Nigeria and Senegal*  
**Aniefiok Udoakang**  
Université Cheikh Anta Diop de Dakar, Senegal
- 06** *Using computational methods to map multigenic disease phenotypes: an ALS toolbox*  
**Styliani Papadaki**  
University of Patras, Patras, Greece
- 07** *Genetic Investigation of South Africans with the Noonan Syndrome Phenotype using Targeted Next Generation Sequencing*  
**Cedrik Ngongang**  
University of Cape Town, Cape Town, South Africa
- 08** *Validation of Cepheid Xpert® BCR-ABL Monitor and Ultra test kits used in the molecular monitoring of chronic myeloid*  
**Chantal De Long**  
Stellenbosch University, Stellenbosch, South Africa
- 09** *Identifying new genes and variants involved in Hearing Impairment in Cameroon, using Next Generation Sequence data*  
**Edmond Wonkam Tingang**  
University of Cape Town, Cape Town, South Africa
- 10** *A case study of X-linked MICPCH caused by a contiguous gene deletion at Xp11.4p11.3*  
**Rizqa Sulaiman-Baradien**  
University of Cape Town, Cape Town, South Africa
- 11** *A report of Williams-Beuren syndrome in a South Africa . . .*  
**Ilse Crous**  
University of Cape Town, Cape Town, South Africa
- 12** *Prothrombin mutation prevalence in hereditary thrombophilia*  
**Shareefa Isaacs**  
Stellenbosch University, Stellenbosch, South Africa

- 13** *The Sudanese Genetic Variation Portal Enabling personalized medicine in understudied populations*  
**Mohammed Omar Elsiddieg Abdallah**  
 University of Khartoum, Sudan
- 12:00-13:30  
**Poster Number**
- 14** *Breast Cancer Risk Perceptions and Genetic Counseling*  
**Derrick Bary Abila**  
 Makerere University, Uganda
- 15** *Ligation detection reaction: the new age for genetic disease research*  
**Priscilla Abena Akyaw**  
 Noguchi Memorial Institute for Medical Research, Ghana
- 16** *A systematic assessment of the Copy Number Variation (CNV) landscape in ADME genes in Sub-Saharan African populations*  
**Laura Cottino**  
 University of the Witwatersrand, Johannesburg, South Africa
- 17** *Web based genomic medicine training for nurses in rural Cameroon: Report, Experience and Impact*  
**Kengne kamga Karen**  
 District Hospital Limbe, Cameroon
- 18** *Sickle Cell Trait and Chronic Kidney Disease*  
**Ernestine Kubi**  
 Noguchi Memorial Institute for Medical Research, Ghana
- 19** *Genetic Architecture of Clinically Relevant Variations*  
**Gerald Mboowa**  
 Makerere University, Uganda
- 20** *A Case report of a novel homozygous splice site mutation in PLA2G6 gene causing Infantile Neuroaxonal dystrophy in a Sudanese family*  
**Melka EL-Amin**  
 The Sudanese, Neurogenetics Group, Sudan
- 21** *Admixture mapping of TB susceptibility in two admixed African populations*  
**Yolandi Swart**  
 Stellenbosch University, Stellenbosch, South Africa

- 22** *Genetic Genealogy - Using Y-STRs for: Surname Studies, Population and Forensic Genetics Based on Males*  
**Surina Singh**  
University of KwaZulu-Natal, KwaZulu-Natal, South Africa
- 23** *Genetic differentiation of bony fish species ...*  
**Nathaniel D. Leesolee**  
National Fisheries and Aquaculture Authority, Liberia
- 24** *Quantitative assessment of double-positive JAK2 and CalR mutations in Myeloproliferative Neoplasms (MNP) at Tygerberg Academic Hospital (TAH)*  
**Megan Cousins**  
Stellenbosch University, Stellenbosch, South Africa
- 25** *The development of a precision laboratory for oncology*  
**Ravnit Grewal**  
South African National Bioinformatics Institute, South Africa
- 26** *Assessing concordance among guidelines for genome-guided therapeutic interventions from different research consortia and regulatory bodies*  
**Stefania Koutsilieri**  
University of Patras, Patras, Greece
- 27** *Understanding the genetic causes of Vitiligo*  
**Nonhlanhla Cynthia Mhlongo**  
University of South Africa, South Africa
- 28** *Interleukin-6 (-174>C and -572G>C) gene promoter polymorphisms, C-reactive protein and glycated haemoglobin as predictor of risk of Type II Diabetes Mellitus in obese and non obese subject*  
**Oyekale Adesola**  
Ladoke Atkintola University of Technology, Ogbomoso, Nigeria
- 29** *Genetics Study of Hearing Loss in Mali: Preliminary Data*  
**Abdoulaye Yalcouye**  
University of Sciences, Techniques and Technology of Bamako, Mali
- 30** *High utility of active tuberculosis case finding in...*  
**Gebeyehu A. Mitku**  
Armauer Hansen Research Institute, Ethiopia

