Autism, epigenetics, pharmacogenetics, iron dysregulation, medical aid reimbursement policy and ethical issues are among the topics selected for discussion at the 10th Applied Genetics Workshop. This event will take place from 30-31 October 2015 with a special focus on the need for a “Learning Healthcare System” to be explored. So much has been accomplished over the last decade that helped us to move from single-gene analysis to microarrays and next generation/exome sequencing! One of the highlights was the registration of the annual workshop as a Short Course at Stellenbosch University entitled “Clinician Training in Pathology-Supported Genetic Testing to be Applicable to the Benefit of Society”. This was done according to the requirements of the South African National Qualifications Framework (NQF). The content focused on life-long learning was approved at NQF level 8. This relates to a scope of knowledge that engages the learner in an area at the forefront of clinical practice. Understanding the methodologies and techniques relevant to genomics and how to apply such knowledge in a clinical context are necessary for effective research translation. Please join us and learn how to critically review genetic information in specialised contexts in order to develop creative responses to ethical issues and obstacles in the path to personalised medicine. This year the workshop is partly sponsored by a UK-South Africa Researcher Links Grant focused on the development of a clinical practice guideline for pathology-supported genetic testing. As part of this initiative a prize of R 20 000 will be awarded to promote research translation in the field of personalised medicine.

**10th Applied Genetics Workshop:** 30 October 2015 from 8h30-16h00

**Strategic Planning & Lecture Session 1:** 31 October 2015 from 9h00-13h00

**VENUE:** Faculty of Medicine and Health Sciences, Teaching Block, Lecture Room 11, Francie van Zijl Drive, University of Stellenbosch, Tygerberg, CAPE TOWN
REGISTRATION DAY 1: 8h30

9h00: Welcome and Introduction - Prof Nico Gey van Pittius, Deputy Dean: Research Faculty of Medicine and Health Sciences, Stellenbosch University

9h30: A combined service and research approach: Maximising beneficence and respect for autonomy in the application of personalised medicine for breast cancer - Dr Nicola Barsdorf, Head of Health Research Ethics, Stellenbosch University

10h15: Epigenetics and the prospect of personalised interventions in the fight against non-communicable diseases - Dr Stuart Raleigh, Head of the Centre for Physical activity and Chronic Disease, University of Northampton, UK

COFFEE & TEA BREAK: 11h00

11h30: Chromosomal microarray (CytoScan): Genomic diagnostics for patients with intellectual disability and autism - Dr Mike Urban, Head of the Clinical Unit, Clinical Genetics and Genetic Counselling, Stellenbosch University and Tygerberg Hospital

12h00: RNA microarray (MammaPrint): Impact on clinical decision-making in South African patients with early stage breast cancer - Dr Heinrich Pohl, Department of Surgery, Stellenbosch University and Tygerberg Hospital

12h30: Application of targeted next generation sequencing (OncoDeep) and microarray tumour profiling in clinical practice - Dr Ettienne Myburgh, Department of Surgery, Stellenbosch University and Panorama Medi-Clinic

LUNCH: 13h00
13h30: Molecular oncology for clinicians - Dr Johann Riedemann, Medical Officer, Karl Bremer Hospital (PhD in Molecular Oncology, University of Oxford, UK)

14h00: Iron in the brain: Not too much and not too little - Dr Ronald van Toorn, Department of Paediatrics and Child Health, Stellenbosch University and Tygerberg Hospital.

14h30: Whole exome sequencing in patients with iron deficiency diagnosed with multiple sclerosis - Prof Susan J van Rensburg, Department of Pathology, Stellenbosch University.

15h00: Molecular genetics and medical schemes: Where to? - Prof Manie de Klerk, General Manager, Metropolitan Health Risk Management

15h45: Closing remarks: Providing a genomics database resource for clinical interpretation - Prof Maritha Kotze, Department of Pathology, Stellenbosch University and Gknownmix

STRATEGIC PLANNING SESSION DAY 2 - Saturday 31 October 2015 9h00-13h00

All stakeholders are invited to form part of a Working Group to be established for the UK-South Africa Researcher Links Initiative approved for funding by the National Research Foundation from October 2015 to March 2016. The first meeting will be in Cape Town immediately after the workshop (31/10/2015), followed by a series of lectures in different regions of South Africa (program to be finalised).

At this first meeting problems related to the incorporation of genomics in clinical practice will be presented by Dr Ettienne Myburgh, who performed an external audit of the Gknownmix database generated at the interface between the laboratory and clinic. Dr Helen Muir and PhD student Nicole van der Merwe will present case studies and the challenges faced as we move from traditional single- to multi-gene testing and next generation/exome sequencing. Based on the use of an ethics approved protocol showing how research influences clinical practice and vice versa, the need for of a "learning healthcare system" will be explored and motivated by Prof Maritha Kotze. Dr Hilmar Lückhoff will explain how evidence generated from the central genomics database is used to design testing algorithms for conditional approval and reimbursement of genomic tests by participating medical schemes in South Africa. Dr Stuart Raleigh will evaluate the potential benefits and risks of pathology-supported genetic testing and
formulate a strategy for possible introduction of genetic screening tests using a similar process in the UK. Joint health outcome studies related to his field of expertise including Sport Medicine will allow for adjustments to be made over time, resulting in continual improvement of genetic testing across the disease spectrum.

An expert panel will be nominated to participate in the above-mentioned lecture series focused on the incorporation of genomics in routine clinical and dietetic practice. Proposals for topics are invited in the form of peer-reviewed research articles published or accepted for publication during the evaluation period. The best idea by a student or intern for research translation with high potential impact on patient care will be awarded a prize of R20 000, to be announced before the end of March 2016. Please email Prof Maritha Kotze at maritha@sun.ac.za for more details.

Cost: R 250 - Please RSVP by 25 October 2015

Registration form available from Janine Cronje, Tel. 021 9389324 jcronje@sun.ac.za

6 Clinical and 2 Ethics points to be applied for

THANK YOU! We thank all referring clinicians and the many patients who gave informed consent for inclusion of their clinical, pathology and genetic information in the Gknowmix Database. This resource is used following an ethically approved protocol for training of students who in turn help to provide the evidence required for reimbursement of comprehensive genomic tests. GeneTalk proactively addresses the need for better communication between scientists, healthcare practitioners and the public by increasing awareness of the role of genetics in health and disease.