



Registration Form: Applied Genetics Short Course

GeneTalk Invitation - 24 October 2014

The Department of Pathology at the Tygerberg Academic Hospital invites you to attend a workshop on Pathology Supported Genetic Testing

SESSION	1: Research translation	2: Reimbursement model	3: Ethics and genomics
TIME	8h30-11h00 <input type="checkbox"/> YES <input type="checkbox"/> NO	11h15-13h00: <input type="checkbox"/> YES <input type="checkbox"/> NO	13h15-16h00 <input type="checkbox"/> YES <input type="checkbox"/> NO
VENUE	Faculty of Medicine and Health Sciences, Teaching Block, Lecture Room 11, Francie van Zijl Drive, University of Stellenbosch, Tygerberg, CAPE TOWN		
COST	<input type="checkbox"/> R250.00 <input type="checkbox"/> R150.00 for students Please register by 23 October 2014 <u>Please use initials and surname as reference for payment</u> Bank: Standard Bank Account holder: University of Stellenbosch Account Number: 07 300 695 5 Branch Code: 05 06 10		

YOUR CONTACT INFORMATION (Please complete one registration form per person)

Initials		Title	Prof	Dr	Mr	Mrs	Ms	
Name								
Surname								
Address								HPCSA Reg No:
								Practice No:
Tel numbers								
Email								
Speciality								
Food preferences?								

5 Clinical and 2 Ethics points approved by the HPCSA

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Secretary: Janine Cronje or Tel: 9389324 or Fax: 0866009434

GeneTalk Workshop program: 24 October 2014

8h30: Registration [order of speakers subject to change]

SESSION 1: Prioritizing the clinical translation of genomic research

Moderator: Prof Tony Bunn

9h00: Welcome and Introduction – Prof Tony Bunn, Programme Director: MRC-PATH Global Health Innovation Accelerator

9h10: Application of personalized genomic medicine using an integrated service and research approach - **Prof Maritha Kotze**, Dept of Pathology, Stellenbosch University

9h30: Commercialisation support from the Medical Research Council - **Rabogajane Busang**, Strategic Health Innovation Partnership (SHIP) Initiative, South African Medical Research Council

10h00: Overview and first results of the breast cancer genomics SHIP project - **Dr Armand Peeters**, SHIP Project manager, Stellenbosch University

10h30: Realising the ideal of personalised therapy in post-menopausal breast cancer patients - **Dr Karin Baatjes**, Department of Surgery, Stellenbosch University

11h00: COFFEE & TEA BREAK

SESSION 2: Implementation of a new oncogenomic reimbursement model

Moderator: Dr Alison September

11h15: Eight years' experience of breast cancer gene profiling (MammaPrint) in South Africa - **Prof Justus Appfelstaedt**, Head of Tygerberg Breast Cancer Clinic, Stellenbosch University

11h45: Discrepancies between IHC, FISH and RNA-based (TargetPrint) assessment of HER2 status: What does it mean for the patient? - **Kathleen Grant**, Department of Biomedical Sciences, Faculty of Health and Wellness, Cape Peninsula University of Technology

12h15: Breast cancer subtyping incorporating microarray analysis (Blueprint): An external database audit - **Dr Etienne Myburgh**, Panorama Hospital and Stellenbosch University

12h45: Impact of gene profiling on treatment decisions: A case study - **Dr Rika Pienaar**, GVI Oncology, Panorama

13h00: LUNCH

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SESSION 3: Ethics and genomics: Balancing the risks and benefits

Moderator: Prof Maritha Kotze

13h15: Metabolic insight into non-communicable diseases: Defining the components of a healthy diet - **Prof Nola Dippenaar**, Formerly Dept of Physiology, University of Pretoria

14h00: Development of referral guidelines for genomic tests using a database resource for translational medicine - **Dr Hilmar Luckhoff**, Dept of Pathology, Stellenbosch University

14h30: Identification of disease-causing gene variants using whole genome/exome sequencing - **Dr Nicki Tiffin**, South African Bioinformatics Institute, University of the Western Cape

14h45: Ethics and informed consent for human genomic research - **Dr Nicki Tiffin**, South African Bioinformatics Institute, University of the Western Cape

15h30: Genomic Research Advancing Clinical Practice (GRACE) initiative - **Dr Frans Cronje**, Stellenbosch University

15h45: Closing remarks – **Prof Manie de Klerk**, Metropolitan Health Group and University of Stellenbosch Business School

PROGRAMME: 9th Applied Genetics Workshop

Session 1 - 9h00-11h00: Prioritizing the clinical translation of genomic research

Extensive genetic research conducted in the diverse South African population over the preceding two decades provided a framework for the development of a clinically integrative approach to personalized genomics. An open-innovation platform was developed to link genetic research with service delivery performed at the interface between the laboratory bench and the bedside. **Prof Maritha Kotze** and **Rabogajane Busang** will discuss how the implementation of a pathology-supported genetic testing (PSGT) model could promote the clinical translation of genomic medicine in relation to appropriate research commercialization. The establishment of a MRC-funded Strategic Health Innovation Partnership (SHIP) focused on breast cancer research aims to facilitate clinical research translation and promote collaboration between academic institutions and the private sector. **Dr Armand Peeters** who was appointed as the SHIP project leader will give an overview and update of the progress, followed by **Dr Karin Baatjes** whose PhD study involving pharmacogenomics is funded as part of this initiative.

Session 2 - 11h15-13h00: Implementation of a new oncogenomic reimbursement model

Oncogenomics is currently leading the way in the field of personalized medicine. **Prof Justus Appfelstaedt** will provide an overview of the clinical implementation of microarray-based gene expression profiling using a 70-gene assay for chemotherapy selection. This MammaPrint test provides an excellent example of how emerging genomic applications could successfully be incorporated into the local healthcare system, as evidenced by its reimbursement by several South African medical schemes. PhD student **Kathleen Grant** and **Dr Etienne Myburgh** will discuss how

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research aimed at validating the performance and clinical relevance of microarray-based assessment of hormone receptor status as well as molecular tumour profiling (provided routinely as an extension of the MammaPrint service) against existing diagnostic standards improves quality assurance and increases confidence in its application aimed at adding value to patient management. **Dr Rika Pienaar** will present a case study of a self-referred breast cancer patient, which highlights the need for greater awareness amongst clinicians of new advanced technologies that can improve the standard of care for breast cancer both locally and abroad.

Session 3 - 13h15-16h00: Ethics and genomics: Balancing the risks and benefits

A number of challenges currently impede the clinical application of genetic testing still largely restricted to a limited number of diagnostic or selected oncogenomic applications. **Prof Nola Dippenaar** will discuss the importance of optimal nutrition to reduce the risk of chronic non-communicable diseases known to have a genetic component. **Dr Hilmar Lückhoff** will explain the value of patient databases linked to the research component of the pathology-supported genetic testing service as a valuable resource for development of pre-screening algorithms that may allow for goal-directed application of genetic testing in eligible individuals. Results of a genetic test should not be used in isolation, but must always be integrated and contextualised within the family history and clinical profile of a patient. The challenges and opportunities associated with exome and whole genome sequencing as the next step in the rapidly developing field of human genetics will be discussed by **Dr Nicki Tiffin**, who recently published together with her co-workers on ethical aspects, biobanking and informed consent for genomic research in Africa. **Dr Frans Cronje** was asked to summarise the insights gained as a result of this workshop in order to define the way forward in line with the SHIP project objectives. The meeting will be closed by **Prof Manie de Klerk** (General Manager MHRM and Associate Professor: USB, Extraordinary lecturer: Faculty of Health Sciences), who proposed a process for funding of genetic testing of common chronic disorders that may soon be reimbursed based on principles derived from the funding of the 70-gene Mammamprint profile.

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.

For more information please contact Prof Maritha Kotze at 021 9389324 / 0828799108 or email maritha@sun.ac.za

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.



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