Genomics on the Move

Workshop Program

7th – 9th September 2015: 08:00-17:00

Room MSL111, First Floor, Mathematical Sciences Laboratory, Science Stadium, West Campus, University of the Witwatersrand, Johannesburg

Hosted by the SBIMB and CRG

TIME	MONDAY 7th September 2015	FACILITATOR
8:00-12:30	**Identification of Mendelian disease variants and cancer driver gene mutations using Exome sequencing** Basic training in the analysis of Next Generation Sequencing data, with a focus on Exome-sequencing analysis for discovery of Mendelian disease variants and mutations in cancer driver genes. ***Introductory Lecture**:* Clinical Applications of Exome Sequencing ***Workshop Part 1*** - Introduction to Mendelian disease case - Introduction to analysis tools - Quality control of Exome-seq data - Sequence alignment - SNP detection - Indel detection - Quality control and filtering of variants - Functional annotation of variants - Candidate gene prioritization - Collecting additional (clinical) information for candidate genes - Overview of useful online resources and tools not used during the course	S Ossowski
12:30-13:30	LUNCH	
13:30-17:00	***Practical Session:*** Data handling and manipulation	S Ossowski
TIME	**TUESDAY 8th September 2015**	**FACILITATOR**
8:00-09:30 09:30-12:30	***Guest Lecture:*** African population genomes and disease variant discovery ***Workshop Part 2*** - Introduction to cancer case - Somatic variant calling - Cancer variant annotation - Cancer databases (Cosmic, IntOGen)	M Ramsay S Ossowski
12:30-13:30	LUNCH	
13:30-17:00	***Application of genomics and genetics in drug discovery and translational medicine***	F Staedtler
TIME	**WEDNESDAY 9th September 2015**	FACILITATOR
8:00-12:30	**Introduction to RNAseq analysis and discovery of long noncoding RNAs** ***Workshop Part 1*** A brief, hands-on introduction to the analysis of gene expression data from RNA sequencing. Processing of raw reads, map them to the genome, and use them to quantify gene expression. If time permits we will use this data to discover long noncoding RNAs amongst these transcripts ***Practical Session:*** Data handling and manipulation	R Johnson
12:30-13:30	LUNCH	
13:30-17:00	**Transcriptomics** An open-discussion format on the topic of how the expression of genes is regulated in the brain in different clinical situations. - Small RNA toxic molecules in neurodegenerative disorders - Targets of miRNAs and brain function - Role of miRNAs in psychiatric disorders	X Estivill

Workshop Facilitators

PROF STEPHAN OSSOWSKI

Genomic and Epigenomic Variation in Disease Group,
Centre for Genomic Regulation, Barcelona, Spain

Stephan Ossowski has obtained a Master in Computer Science from the University of Tübingen in 2004 and a PhD in Computational Biology from the Max-Planck Institute Germany in 2010. After a short postdoc at the MIT in Boston he has started the Genomic and Epigenomic Variation in Disease group at the Centre for Genomic Regulation in Barcelona. Stephan's expertise lies in computational genomics and epigenomics, genetic diseases and cancer. In 2008 he published the first whole-genome analysis of a plant genome using next generation sequencing technology and has since worked on developing novel computational and statistical methods for NGS analysis.

PROF MICHÈLE RAMSAY

Sydney Brenner Institute for Molecular Bioscience,
Department of Human Genetics, Faculty of Health Sciences,
University of the Witwatersrand, Johannesburg

Michèle Ramsay is Director of the Sydney Brenner Institute for Molecular Bioscience (SBIMB) and a Professor in the Division of Human Genetics, University of the Witwatersrand, Johannesburg. Her research interests include African population genetic and epigenetic diversity and their role in diseases exacerbated by adverse lifestyle choices, including obesity and cardiometabolic diseases. She collaborates on genetic research into eye diseases and autoimmune diseases in African populations and studies the role of epigenetic changes in a mouse model for fetal alcohol spectrum disorders (FASD). Michele is a member of the Human Heredity and Health in Africa Consortium.

DR FRANK STAEDTLER

Novartis Institutes of Biomedical Research,
Novartis, Basel, Switzerland

Frank Staedtler is a molecular geneticist and biologist experienced in creating and leading genomics/ genetics laboratories as well as cross-functional biomarker projects in pharmaceutical R & D. In a recent role at the Novartis Institutes of Biomedical Research (NIBR), Switzerland he built and headed a cluster of four laboratories called Applied Human Genetic and Genomic Technologies in Biomarker Development/Translational Medicine. In this role he was also fostering scientific exchange and cross-cultural experience in working with South African scientists.

Frank's passion is to enable precision medicine by enhancing our understanding of diseases and how drugs work with regard to their safety and efficacy. This is done through the application of biomarker science and leading genomics and genetics technologies. Prior to this Frank spent his Postdoc

implementing transgenic safety models at Sandoz Pharma, Toxicology. During this time he received a post-graduate degree in toxicology. Before the period in industry, Frank earned his diploma and doctoral degree on basic genetic research topics at the Max-Planck-Institute in Cologne, and the University in Bonn, Germany.

DR RORY JOHNSON

Computational Biology of RNA Processing Group,
Centre for Genomic Regulation, Barcelona, Spain

Rory studied physics and biophysics at Imperial College London. He won a Wellcome Trust scholarship to complete his PhD at the University of Leeds, studying the regulation of coding and non-coding genes in the nervous system. During his postdoc at the Genome Institute of Singapore, he applied next generation sequencing approaches to mapping gene regulatory networks in embryonic stem cells. Since 2010 he has been a Ramon y Cajal fellow at the Centre for Genomic Regulation, participating in a number of experimental and bioinformatic projects to understand the function of long non-coding RNAs.

PROF XAVIER ESTIVILL

Genomics and Disease Group
Centre for Genomic Regulation, Barcelona, Spain

Xavier Estivill graduated in Medicine, specialised in Haematology, and obtained his doctorate in Medicine (MD) by the Autonomous University of Barcelona and in Philosophy (PhD) by the University of London. He is currently Senior Group Leader at the Centre Genomic Regulation (CRG), Associate Professor of the Pompeu Fabra University (UPF), and Director of Genomics and Personalized Medicine Unit at Dexeus Woman's Health in Barcelona.

His research groups have contributed to the understanding of cystic fibrosis, he defined the spectrum of the molecular pathology and history of mutations in the European population, and discovered the role of CFTR mutations in male infertility. His group demonstrated germline mosaicism for this neurofibromatosis, and discovered the cellular origin of the NF1 mutations. In Down syndrome, he has discovered genes with important roles in neurogenesis, synaptic regulation and control of transcription. His group identified some of the major genes and mutations that cause hereditary deafness, detected nucleotide and structural variants involved in inflammatory disorder and psychiatric diseases. He has published over 600 research papers and his group has received numerous awards.

Recent achievements of his group have been on the identification of genetic variants associated with psoriasis and several psychiatric diseases, the description of toxicity of small RNAs in Huntington's disease and the identification of novel small RNA molecules in the human genome. He is promoting activities of translational genomics based on a multilayer complete characterization of the individual genome. He currently coordinates the germline mutations group of the International Cancer Genomics Consortium on PanCancer analysis.