IX CRG ANNUAL SYMPOSIUM
28/29 October 2010, Barcelona (Spain)
Medical Genome Sequencing:
Understanding the Genomes of Disease
Organizers: Xavier Estivill & Roderic Guigó

CRG Centre for Genomic Regulation

Thursday 28 October – Defining the Framework of Genomic Analysis

8:00 – 9:00 Registration

9:00 – 9:05 Welcome by Miguel Beato

9:05 – 9:10 Introduction by Xavier Estivill

Chair: Stephan Ossowski (Center for Genomic Regulation (CRG), Spain)

9:10 – 9:50 **Michael Metzker** (Human Genome Sequencing Center, Baylor College of Medicine, USA)
Next generation technologies – Basics and applications

9:50 – 10:30 **Ivo Gut** (National Centre of Genomic Analyses, Spain)
Applications of whole-genome sequencing

10:30 – 10:50 **Mario Cáceres** (Biotechnology and Biomedicine Institute UAB, Spain)
Bioinformatic prediction of non-redundant polymorphic inversions in the human genome

10:50 – 11:20 **Coffee Break**

11:20 – 12:00 **Ann-Christine Syvanen** (Uppsala University, Sweden)
Allele-specific gene expression as a guide to genes with cis-acting regulatory epigenetic and genetic factors

12:00 – 12:40 **Jun Wang** (Beijing Genomics Institute, China & University of Copenhagen, Denmark)
Personal genomes are personalized

12:40 – 13:00 **Luca Pagani** (Wellcome Trust Sanger Institute, UK)
Characterization, through re-sequencing, of genetic variants associated with high altitude adaptation in North Caucasians

13:00 – 14:30 **Lunch and Poster Session**

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Chair: Fyodor Kondrashov (Center for Genomic Regulation (CRG), Spain)

14:30 – 15:10 **Jeffrey Barrett** (Wellcome Trust Sanger Institute, UK)
The case of the missing heritability: clues so far and mysteries remaining

15:10 – 15:50 **Paul Flicek** (European Bioinformatics Institute, UK)
Annotating and understanding human variation

15:50 – 16:10 **Victor Solovyev** (University of London, UK)
A tool for reconstructing sequences and transcriptome analysis using next-generation sequencing data

16:10 – 16:40 **Coffee Break**

16:40 – 17:20 **Xavier Estivill** (Center for Genomic Regulation (CRG-UPF), Spain)
Structural variation analysis by large-scale human genome sequencing

17:20 – 18:00 **Jonas Korlach** (Pacific Biosciences, USA)
Applications of Single-Molecule, Real-Time (SMRT™) DNA sequencing

18:00 – 18:40 **Steve Lincoln** (Complete Genomics, USA)
Large-scale human genome sequencing service for advanced disease studies

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Friday 29 October – Genomic Dissection of Phenotypes

8:00 – 9:00 Registration

Chair: Núria López-Bigas (Universitat Pompeu Fabra (UPF), Spain)

9:00 – 9:40 **Emmanouil T. Dermitzakis** (University of Geneva, Switzerland)
Cellular population genomics in humans

9:40 – 10:20 **Jennifer Meadows** (Dept of Medical Biochemistry and Microbiology, Uppsala University, Sweden)
The power of comparative genetics and genomics for finding genes of medical relevance

10:20 – 10:40 **Mireia Jordà** (Institute of Predictive Medicine and Personalized Cancer Medicine, Spain)
Filling up gaps in epigenomic maps: the active Aluome

10:40 – 11:10 **Coffee Break**

11:10 – 11:50 **Sarah Ng** (Genome Sciences & Pediatrics, University of Washington, USA)
Next-generation Mendelian genetics by exome wequencing

11:50 – 12:30 **Joris Veltman** (Radboud University, Nijmegen Medical Centre, The Netherlands)
Next generation sequencing in the clinic

12:30 – 12:50 **Raquel Rabionet** (Center for Genomic Regulation (CRG-UPF), Spain)
Using homozygosity mapping and exome sequencing to search for a gene causing a rare articular disease

13:00 – 14:30 **Lunch and Poster Session**

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Chair: Cedric Notredame (Center for Genomic Regulation (CRG), Spain)

14:30 – 15:10 **Stephan Schreiber** (Institute of Clinical & Molecular Biology, Germany)
Crohn disease, paradigm for the etiology of complex inflammatory disorders

15:10 – 15:50 **Roderic Guigó** (Center for Genomic Regulation (CRG-UPF), Spain)
Uncovering and understanding splicing through massively parallel sequencing

15:50 – 16:10 **Yu Sun** (Leiden University Medical Center, The Netherlands)
Terminal osseous dysplasia is caused by a single recurrent mutation in the FLNA gene

16:10 – 16:40 **Coffee Break**

16:40 – 17:20 **Stylianos Antonarakis** (University of Geneva, Switzerland)
Exome re-sequencing of seven melanoma cell lines to characterize somatic mutations

17:20 – 18:00 **Peter Campbell** (Wellcome Trust Sanger Institute, UK)
Interrogating the architecture of cancer genomes

18:00 – 18:40 **Manolis Kellis** (Massachussets Institute of Technology and Broad Institute, USA)
Disease epigenomics: Interpreting disease-associated non-coding variants using chromatin states and activity profiles across many human ENCODE cell types

18:40 – 18:50 Closing remarks by Roderic Guigó