



# EXOME SEQUENCING ANALYSIS IN DISEASE & CLINICAL RESEARCH

CRG, Barcelona, 25-28 November, 2013

## PRELIMINARY SCHEDULE

### Monday November 25

16:00-17:00 *Introductory Lecture*

Stephan Ossowski: *Clinical Applications of Exome Sequencing*

17:00-20:00 *Practical work*

- Introduction of two clinical example cases
- Introduction to basic computational analysis tools
- Inspecting Exome-seq result files
- Quality control of Exome-seq data

### Tuesday November 26

16:00-17:00 *Introductory Lecture*

Oliver Drechsel: *Computational Analysis of Exome-seq Data*

17:00-20:00 *Practical work*

- In-depth discussion of use case 1: parent-child trio
- Sequence alignment
- SNP detection

### Wednesday November 27

16:00-17:00 *Lecture*

Xavier Estivill: *Next Generation Diagnostics of Metabolic Disorders by Targeted Sequencing*

17:00-20:00 *Practical work*

- In-depth discussion of use case 2: familial hyperkalemic hypertension
- Functional analysis of SNPs
- Collecting additional information for genes affected by SNPs



**Thursday November 28**

*16:00-17:30 Lectures*

Sergi Beltran Agulló: TBA

Stephan Ossowski: *Identification of Driver and Susceptibility Genes in the ICGC Chronic Lymphocytic Leukemia Cohort*

*17:30-20:00 Practical work*

- Prioritization of causal variants and target genes
- Web (e.g. OMIM) and literature (e.g. Pubmed) search of potential target genes