

# **Finding genes for human disease**

from genetics to genomics

Oporto  
15 - 19 April 2013

## **Faculty**

Marianna Bevova, Peter Heutink, Yurii Aulchenko

# Program

## **Monday 15<sup>th</sup> April (Room B)**

9:15-10:00	Lecture 1:	Variation in human genome (Peter Heutink)
10:15-11:00	Lecture 2:	Monogenic disorders. Linkage analysis, Next generation sequence approach (Marianna Bevova)
11:30-13:30	Exercise:	Analysis of the next generation sequence data (Marianna Bevova)
13:30-14:30	Lunch	
14:30-18:00	Exercise:	Analysis of the next generation sequence data (Marianna Bevova)

## **Tuesday 16<sup>th</sup> April (Room B)**

9:15-10:00	Lecture 1:	Functional studies. From Bioinformatics to Practice (Peter Heutink)
10:30-13:30	Exercise:	Virtual Cloning (haplotype analysis) (Marianna Bevova)
13:30-14:30	Lunch	
14:30-15:30	Lecture 2:	Multifactorial disease and association studies (Yurii Aulchenko)
15:30-18:00	Exercise:	Introduction to R and association analysis (Yurii Aulchenko)

## **Wednesday 17<sup>th</sup> April (Room B)**

9:15-10:00	Lecture 1:	Genome wide association studies (Yurii Aulchenko)
10:15-11:00	Lecture 2:	Power and coverage of genome wide association studies (Yurii Aulchenko)
11:30-13:30	Exercise:	Power estimation; GWAS QC
13:30-14:30	Lunch	
14:30-15:30	Lecture 3:	Confounding in Genome wide association studies (Yurii Aulchenko)
15:30-16:30	Exercise:	GWAS in GenABEL (Yurii Aulchenko)
17:00-18:00	Exercise:	Developing a study design

**Thursday 18<sup>th</sup> April (Room C)**

9:15-10:00	Lecture 1:	Rare variants high throughput sequencing. Introduction. (Marianna Bevova)
10:15-11:00	Lecture 2:	Analysis of the rare variants (Yurii Aulchenko)
11:15- 12:00	Lecture 3:	Genetic Interactions (Yurii Aulchenko)
12:30-13:30	Exercise:	Developing a study design
13:30-14:30	Lunch	
14:30-18:00	Exercise:	Developing a study design

**Friday 19<sup>th</sup> April (Morning: Room B & Afternoon: Aud. A)**

9:15-10:00	Lecture 1	Positional cloning for complex diseases/functional annotation (FANTOM, ENCODE) (Peter Heutink)
10:15-11:00	Lecture 2:	Applications of the medical genetics findings (diagnostic, pharmacogenomics) (Marianna Bevova)
11:30-13:30	Exercise:	Developing a study design
13:30-14:30	Lunch	
14:30-18:00		Presentation and discussion of the study design