

# **Finding genes for human disease**

from genetics to genomics

Oporto

7 - 11 July 2014

## **Faculty**

Marianna Bevova, Michel Georges, Yurii Aulchenko

# Program

## Monday 7<sup>th</sup> July

9:30-9:45	Course introduction (Marianna Bevova)
9:45-10:30	Lecture: Variation in human genome (Marianna Bevova)
10:45-11:30	Lecture: Monogenic disorders. Linkage analysis (Marianna Bevova)
11:45-13:30	Exercise: Lod score exercise (Marianna Bevova)
13:30-14:30	Lunch
14:30-15:30	Lecture: Monogenic disorders. Next generation sequencing (Marianna Bevova)
15:45- 18:00	Exercise: Virtual Cloning (haplotype analysis)

## Tuesday 8<sup>th</sup> July

9:30-10:00	Discussion Exercises (Marianna Bevova)
10:00-13:30	Exercise: Analysis of the next generation sequence data (Marianna Bevova)
13:30-14:30	Lunch
14:30-15:30	Lecture: Multifactorial disease and association studies (Yurii Aulchenko)
15:30-18:00	Exercise: Introduction to R and association analysis (Yurii Aulchenko)

## Wednesday 9<sup>th</sup> July

9:30-10:00	Lecture: Genome wide association studies (Yurii Aulchenko)
10:15-11:00	Lecture: 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: 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 10<sup>th</sup> July**

9:30-10:15	Lecture:	Positional cloning for complex diseases (Michel Georges)
10:30-11:15	Lecture:	Functional annotation of the human genome (Michel Georges)
11:30-13:30	Exercise:	Developing a study design
13:30-14:30	Lunch	
14:30-18:00	Exercise:	Developing a study design

### **Friday 11<sup>th</sup> July**

9:30-10:15	Lecture:	Applications of the medical genetics findings (diagnostic, pharmacogenomics) (Marianna Bevova, Yurii Aulchenko)
10:30-11:15	Lecture:	Beyond exome sequencing and GWAs: Functional Genomics
1:15-13:30	Exercise:	Developing a study design
13:30-14:30	Lunch	
14:30-17:00		Presentation and discussion of the study design. Conclusions