# Finding genes for human disease

## from genetics to genomics

## Porto

## 16 – 20 May 2016

### Faculty

Olga Sin, Yurii Aulchenko, Marianna Bevova

# Program

**Monday 16<sup>th</sup> May**

 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- 17:00           Exercise:        Virtual Cloning (haplotype analysis)


**Tuesday 17<sup>th</sup> May**

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)

15:30-17:00           Exercise:        Introduction to R and association analysis
(Yurii Aulchenko)


**Wednesday 18<sup>th</sup> May**

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           Exercise:        GWAS in GenABEL
(Yurii Aulchenko)

15:30-17:00           Exercise:        Developing a study design

**Thursday 19th May**

| | | |
|---|---|---|
| 9:30 -10:15 | Lecture: | Functional Genomics: from gene to function (Olga Sin) |
| 10:30 -11:15 | Lecture: | Finding modifiers of protein aggregation in neurodegenerative diseases (Olga Sin) |
| 11:30-13:30 | Exercise: | Developing a study design |
| 13:30-14:30 | Lunch | |
| 14:30-17:00 | Exercise: | Developing a study design |

**Friday 20th May**

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|---|---|---|
| 9:30-10:15 | Lecture: | Applications of the medical genetics findings (diagnostic, pharmacogenomics) (Marianna Bevova) |
| 10:30 -13:00 | Exercise: | Developing a study design |
| 13:00-14:00 | Lunch | |
| 14:00-17:00 | | Presentations and discussion of the study design. Conclusions |