MEDICAL GENETICS

Course Workload		Assessment form (examination/ graded test/ ungraded test)
ECTS	Hours	
6	216	Exam

The course deals with the issues of working with human genomic data: both from the point of view of a genome-wide search for associations, and from the point of view of interpretation of exome data.

Course structure:

1. HUMAN GENETICS FOUNDATIONS
    1.1. Mendel and post-Mendel era.
    1.2. Pre-genome era. Mapping of the first human disease gene. Huntingnton's disease.

2. GENE MAPPING AND DNA SEQUENCING
    2.1. RFPL, microsatellite and genetic linkage.
    2.2. Pedigree and linkage analysis.
    2.3. DNA forensics.
    2.4. Human Genome Project. SNP map of the human genome. DNA variation.
    2.5. Linkage disequilibrium and genome-wide association studies. HapMap project.

3. GWAS STUDIES
    3.1. GWAS concepts and approaches.
    3.2. Hands on GWAS tutorial.
    3.3. GWAS discussion and resources.
    3.4. Polygenic risk scores.
    3.5. UK biobank.

4. OTHER GENETIC STUDIES AND RESOURCES
    4.1. Next generation sequencing.
    4.2. Exome sequencing.
    4.3. GATK pipeline.
    4.4. Variant annotations, selection pressure metrics.
    4.5. Large scale sequencing resources (ExAC & GnomAD).
    4.6. Rare variant association studies.
    4.7. Hands-on analysis of exome sequencing data.