האוניברסיטה העברית בירושלים THE HEBREW UNIVERSITY OF JERUSALEM



Syllabus

Personal Genomics - 52634

Last update 27-09-2017

HU Credits: 3

**Responsible Department:** statistics

Academic year: 0

Semester: 1st Semester

Teaching Languages: Hebrew

Campus: Mt. Scopus

Course/Module Coordinator: Or Zuk

<u>Coordinator Email: or.zuk@mail.huji.ac.il</u>

Coordinator Office Hours: Mondays 12-13

## Teaching Staff: Dr. Or Zuk

Course/Module description: The course will introduce the major methods for analyzing and interpreting personal genomes.

We will focus on the statistical models and computational tools used in the field. There will be theoretical and computational exercises requiring basic knowledge of *R*/python.

<u>Course/Module aims:</u> To teach the basic models used in personal genomics

## Learning outcomes - On successful completion of this module, students should be able to:

To use basic tools for interpreting personal genomes

Attendance requirements(%):

0

Teaching arrangement and method of instruction: lectures

Course/Module Content:

1. Basic introduction to genetics and mutational models.

*(i) Genome organization, DNA, chromosomes, mitosis&meiosis: mutations, recombination* 

*(ii) Eukariotic Gene structure: exons, introns, transcription, translation: genetic code* 

2. Quantitative and Population Genetics:

(i) Pedigrees, Kinship

(ii) The Fisher-Wright model and its extensions

(iii) Coalescent theory: gene genealogies, recombination,

(iv) Demography: Ancestry, population structure

(v) Phasing and Imputation

3. Massively Parallel Sequencing Technologies:

(i) Short read alignment, variant calling

(ii) chip-seq/rna-seq - sequencing for quantification

4. Complex Traits:

*(i) Genome Wide Association Studies* 

(ii) Risk Prediction, Missing Heritability, common and rare variants

(iii) Prediction of variants severity

*(iv) Scalable GWAS to large samples* 

5. Measures and tests for natural selection:
(i) Types of selection (purifying, positive, balancing).
(ii) Descriptive statistics: F\_st, Tajima's D
(iii) Tests based on haplotype length and site-frequency specturm
(iv) Inter-species tests: DN/DS, HKA, Mcdonald-Kreitman

<u>Required Reading:</u> None

<u>Additional Reading Material:</u> 1. Coalescent Theory: An Introduction - J. Wakeley

- 2. Mathematical Population Genetics 1 W. Ewens
- 3. Genetics and Analysis of Quantitative Traits Lynch&Walch

Course/Module evaluation: End of year written/oral examination 0 % Presentation 0 % Participation in Tutorials 0 % Project work 0 % Assignments 60 % Reports 0 % Research project 0 % Quizzes 0 % Other 40 % Home exam

Additional information: