

## INTEGRATED APPLIED GENETICS TRAINING - AppGENEdu

*A Project Financed through the EEA Grants 2014-2021, Education, Scholarships, Apprenticeships and Youth Entrepreneurship Programme in Romania, Cooperation projects in The Higher Education Area*

### Summer School Programme

#### Week 1

##### Day 1

Students Welcome

Genetics perspective – Prof. Bohiltea L

Epidemiology perspective - Conf. Pitigoi D

Genetics Fundamentals I – Bohiltea L, Radoi V, Ursu R

Genetics Fundamentals II – Bohiltea L, Radoi V, Ursu R

##### Day 2

Statistics and Bioinformatics - Manolescu A, Poenaru E

- Fundamental elements of biostatistics
- Fundamental elements of statistical genetics
- Elements of statistical study design

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Workshop – Poenaru E, Dogaru C, Iordache P, Poenaru C

- Statistical tools; R
- Statistical basic processing

Workshop – Poenaru E, Dogaru C, Iordache P, Poenaru C

- Statistical basic processing
- Basic data visualization

##### Day3

Fundamentals of Genetic Epidemiology – Pitigoi D, Manolescu A

- Quantitative Statistical Tests, linear regression (smoking)
- Phenotype - genotype correlations

Fundamentals of Genetic Epidemiology – Pitigoi D, Manolescu A

- Workshop – Iordache P, Poenaru E, Manolescu A, Poenaru C

- Phenotype - genotype correlations
- Example of epidemiology study design based on provided "use-cases"

Workshop – lordache P, Poenaru E, Manolescu A, Poenaru C

- Phenotype - genotype correlation
- Example of epidemiology study design based on provided "use-cases"

## Day3

Elements of genetic risk analysis, Part I – Manolescu A, lordache P, Ursu R, Radoi V

- Genetics: LD frequencies, HWE
- Relative risk

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- Genetics: LD frequencies, HWE
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Workshop biostatistical tools - plink – lordache P, Ursu R, Radoi V, Poenaru E, Manolescu A, Poenaru C

- Missingness by phenotype
- Missingness by genotype
- Hardy-Weinberg
- Allele frequencies
- LD-based SNP pruning
- Mendel errors

Workshop biostatistical tools - plink – lordache P, Ursu R, Radoi V, Poenaru E, Manolescu A, Poenaru C

- Missingness by phenotype
- Missingness by genotype
- Hardy-Weinberg
- Allele frequencies
- LD-based SNP pruning
- Mendel errors

## Day5

Elements of genetic risk analysis, Part II – Manolescu A, lordache P, Ursu R, Radoi V

- Genetics: linkage disequilibrium, haplotypes
- Example of population admixture

Bioinformatics methods & principles

- Elements of genetic risk analysis, Part II – Manolescu A, lordache P, Ursu R, Radoi V
- Genetics: LD frequencies, HWE

Workshop – lordache P, Ursu R, Radoi V, Poenaru E, Manolescu A, Poenaru C

- Case/control
- Fisher's exact
- Stratified analysis
- Quantitative trait
- Linear and logistic models
- Multiple-test correction

Workshop – lordache P, Ursu R, Radoi V, Poenaru E, Manolescu A, Poenaru C

- Missingness by phenotype
- Missingness by genotype
- Hardy-Weinberg
- Allele frequencies
- LD-based SNP pruning
- Mendel errors

## Week 2

### Day 6

Elements of Data Science – Poenaru C, Iordache P, Poenaru A

- Data load
- Data cleansing
- Data visualization
- Data processing
- Data comparison
- Reproducible research

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Workshop – Poenaru C, Iordache P, Poenaru E, Dogaru C

- Example of data processing (from problem to result)
- Bioinformatics exercises

Workshop – Poenaru C, Iordache P, Poenaru E, Dogaru C

- Example of data processing (from problem to result)
- Bioinformatics exercises

### Day 7

Bioinformatics - Iordache P, Ursu R, Radoi V

- Biologic consequences of genetic variants/gene mutations
- Genetic variances in Genetic Epidemiology context (GWAS)

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- Biologic consequences of genetic variants/gene mutations
- Genetic variances in Genetic Epidemiology context (GWAS)

Workshop – Iordache P, Ursu R, Radoi V, Poenaru E

- GWAS study
- Output analysis

Workshop – Iordache P, Ursu R, Radoi V, Poenaru E

- GWAS study
- Output analysis

### Day 8

Advanced Applied Genetics – Ursu R, Radoi V, Iordache P

- Replication studies
- Quantitative trait loci
- Genetic pathways & gene integration

Advanced Applied Genetics – Ursu R, Radoi V, Iordache P

- Replication studies
- Quantitative trait loci
- Genetic pathways & gene integration

Workshop – Iordache P, Ursu R, Radoi V

- Replication study
- Visual representation

Workshop – Iordache P, Ursu R, Radoi V

- Replication study
- Visual representation

## Day 9

Bioethics principles – Curca C, Chirica V

- Informed consent
- GDPR elements
- Elements of ethics of scientific publication

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Elements of Research methodology – Halldorsson B, Mates D, Manolescu A, Iordache P,  
Specific steps and examples for genomics data science projects

Specific steps and examples for genomics data science projects – Halldorsson B, Mates D,  
Manolescu A, Iordache P

## Day 10

Prepare and writing a scientific article – Vinereanu D

Project bid and presentations

## Week 3

## Day 11

Visits to the clinics associated with the project topic – Jinga V, Rascu S, Sima C, Jinga M,  
Vasile D, Vinereanu D, Trasca L, Mehedinți A

- Phenotypic and epidemiological data collection
- Genetic sampling
- Clinical visit
- Clinical case report

## Day 12

Visits to the clinics associated with the project topic – Jinga V, Rascu S, Sima C, Jinga M,  
Vasile D, Vinereanu D, Trasca L, Mehedinți A

- Phenotypic and epidemiological data collection
- Genetic sampling
- Clinical visit
- Clinical case report

Project work

## Day 13

Project work

## Day 14

Project work

## Day 15

Project presentation and evaluation