BIO-373

Genetics and genomics

Deplancke Bart, Fellay Jacques

Cursus	Sem.	Type
Life Sciences Engineering	BA5	Opt.
Statistics	MA1, MA3	Opt.

Language of teaching	English
Credits	4
Session	Winter
Semester	Fall
Exam	Written
Workload	120h
Weeks	14
Hours	**4 weekly**
Courses	2 weekly
Exercises	1 weekly
Project	1 weekly
Number of positions	

Summary

The theoretical part of this course covers classical genetics and contemporary genomics. Because bioinformatics has become important for genomic research, the course also includes practical applications to genomic analyses using Python, including group projects.

Content

- Normal and abnormal chromosomes; major chromosomal diseases.
- Different modes of transmission: Mendelian, non-Mendelian, risk factors.
- Importance and limitations of genetic analyses.
- Different types of genetic variants and effect on the individual and the population.
- Examples of the most frequent hereditary diseases, genotype-phenotype correlation.
- What does it mean to be genetically different? What are genomic variants and how can they impact phenotypes?
- Ethical guidelines on genetic research and gene therapy.
- Composition and organization of the genome
- Regulatory networks: what are their components, their architecture and how do they work?
- How is the genome structured in the nucleus of the cell; what is the impact of structural features on the function of the genome?
- Overview of high-throughput sequencing technologies
- How are regulatory networks fine-tuned, and what are the current methodological challenges?
- What is a minimal genome? How can the genome be modified for biotechnological applications?
- What is the future of genomics in the context of personalized medical applications?

Learning Outcomes

By the end of the course, the student must be able to:

- Elaborate on the social and ethical implications of genetics and genomics, including genetic testing and gene therapy
- Assess / Evaluate the differences between mutations, risk factors and genetic variations
- Examine the basics of population and quantitative genetics, evolutionary and conservation genetics
- Describe the architecture of a genome and its function with a specific focus on creating a knowledge base of how the genome interacts with the proteome
- Analyze the structural and functional properties of gene regulatory networks and how these networks coordinate differential gene expression

- Explain how solving the human genome sequence is paving the way for personalized medicine
- Perform basic genomic analyses using Python (differential expression, association study, etc.)

Teaching methods

Ex cathedra, exercices and group project

Expected student activities

Exercises + group project in Python

Assessment methods

Written exam + report on a project in Python

Supervision

Office hours	No
Assistants	Yes
Forum	Yes

Resources

Virtual desktop infrastructure (VDI)

No

Bibliography

Concepts of Genetics / Klug - 10th edition, ISBN 978-0-321-72412-0
Genomes / Brown - 3rd Edition ISBN 0 8153 4138 5

Ressources en bibliothèque

- Genomes / Browne, CRC Press
- Concepts of Genetics / Klug

Moodle Link

- https://go.epfl.ch/BIO-373