



Course syllabus for

Molecular Genetics and Genomics, 5 credits

Molekylär genetik och genomik, 5 hp

This course syllabus is valid from autumn 2023.

Course code	5MT010
Course name	Molecular Genetics and Genomics
Credits	5 credits
Form of Education	Higher Education, study regulation 2007
Main field of study	Molecular Life Science
Level	AV - Second cycle
Grading scale	Pass with distinction, Pass, Fail
Department	Department of Molecular Medicine and Surgery
Decided by	Programme committee for study programmes in biomedicine
Decision date	2023-03-07
Course syllabus valid from	Autumn 2023

Specific entry requirements

A Bachelor's degree or a professional degree worth at least 180 credits. At least 10 credits should be in mathematics and at least 20 credits in life sciences (such as cell biology, biochemistry, microbiology, gene technology, or molecular biology). Proficiency in English equivalent to English B/English 6.

Objectives

The aim of this course is to impart a foundational understanding of human genetics, including the organization and function of the human genome, and to introduce advanced genomic technologies. It also aims to provide students with an understanding of genetic disease mechanisms and the ability to critically evaluate and present genetic research in oral and written form.

Upon completion of the course, the student should be able to:

Regarding knowledge and understanding

- Explain the structure and function of genes and genomes, including how the human genome is organised, how gene expression is regulated, and how genetic variation differs between individuals and species,
- Explain the molecular mechanisms of genetic variation and predict the potential effects of various genetic changes on gene function and phenotype,
- Relate genetic variants to the development of human disease and describe the patterns of inheritance associated with different types of genetic disease.

Regarding competence and skills

- Select appropriate genomic technologies and data analysis strategies to identify diseasecausing genes and explain the rationale behind the selection,
- Evaluate candidate variants and genes using publicly available databases and tools,
- Extract genomic data from publicly available databases,
- Search for, collect, evaluate, interpret, and discuss relevant scientific literature and research findings related to topics within the course.

Regarding judgement and approach

- Reflect on research involving human and animal material from the perspectives of ethics and equal opportunities and describe ways to ensure fair and ethical research practices,
- Evaluate topics within the course from the perspectives of global health and the United Nations Sustainable Developmental Goals (SDGs).

Content

Throughout the course, students will receive an introduction to genetic concepts and the latest genome technologies, with a focus on human genetics in the life sciences. The course will cover a range of themes, including basic principles of the gene structure, gene expression and regulation, genome organization, origin and consequences of genetic variation, basics of inheritance, current genomic technologies and approaches to identify diseasecausing variants in alignment with the course objectives. The course includes selfstudy, lectures, workshops, group work, and oral presentations.

Teaching methods

The learning experience in this course will be active and involve integrated feedback. The course includes "flipped classroom" learning, in which course material is provided before teachermoderated discussions in workshops or seminars. The learning activities for this course will include seminars, lectures, and prerecorded video lectures as part of "flipped classroom" instruction, as well as workshops. Particular emphasis will be placed on peer learning and selfstudy in both group and individual settings.

Examination

The examination consists of assessment of oral presentation (graded Fail/Pass/Pass with distinction) and written assignments (graded Fail/Pass/Pass with distinction). To pass the whole course the grade "Pass" must have been obtained for both examination parts of the course. To attain the grade "Pass with distinction" for the whole course, a grade of "Pass with distinction" must have been obtained for both examination parts of the course.

Students must complete compulsory assignments to pass the course.

Students that fail to submit compulsory assignments in due time before the deadline will lose the opportunity to be graded with "Pass with distinction" for the course.

If there are special grounds, or a need for adaptation for a student with a disability, the examiner may decide to deviate from the syllabus's regulations on the examination form, the number of examination opportunities, the possibility of supplementation or exemptions from the compulsory sections of the course etc. Content and learning outcomes as well as the level of expected skills, knowledge and abilities may not be changed, removed or reduced.

Compulsory participation

The introduction to the course, individual assignments, workshops, and group assignments as well as lectures linked to these parts are compulsory. The course director assesses if and, in that case, how

absence can be compensated. Before the student has participated in all compulsory parts or compensated absence in accordance with the course director's instructions, the student's results will not be registered in LADOK. Absence from a compulsory activity may result in that the student cannot compensate the absence until the next time the course is given.

Limited number of examinations or practical training sessions

Students who have not passed the regular examination are entitled to participate in five more examinations. If the student is not approved after four examinations, he/she is recommended to retake the course at the next regular course date, and may, after that, participate in two more examinations. If the student has failed six examinations/tests, no additional examination or new admission is provided.

The number of times that the student has participated in one and the same examination is regarded as an examination session. Submission of a blank examination is regarded as an examination. An examination, for which the student registered but not participated in, will not be counted as an examination.

Other directives

The course language is English.

Literature and other teaching aids

Latest review articles from high impact journals in the field of genetics and genomics. The articles will be provided during the course.

Strachan, Tom; Read, Andrew P.

Human molecular genetics

Fifth edition. : Boca Raton, Florida : CRC Press, 2019 - xiii, 770 pages

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