GHENT UNIVERSITY

Course Specifications

Valid as from the academic year 2024-2025

Advanced Human Genetics (D012701)

Course size *(nominal values; actual values may depend on programme)*

Credits 6.0 **Study time 180 h**

Course offerings in academic year 2024-2025

A (semester 2)	English	Gent

Lecturers in academic year 2024-2025

Symoens, Sofie	GE31	lecturer-in-charge
Laukens, Debby	GE35	co-lecturer
Syx, Delfien	GE31	co-lecturer
Vanakker, Olivier	GE31	co-lecturer
Vanlander, Arnaud	GE35	co-lecturer

Offered in the following programmes in 2024-2025	crdts	offering
Master of Science in Bioinformatics(main subject Systems Biology)	6	A
Master of Science in Biomedical Sciences	6	A

Teaching languages

English

Keywords

Epigenetics, imprinting, uniparental disomy, haplo-insufficiency, dominant negative, gain-of-function, triplet repeat, GWAS studies, single nucleotide polymorphisms, non-mendelian inheritance, pharmacogenetics, gene-therapy, preimplantation genetic testing.

Position of the course

In this course we will focus on new insights in the genetic basis of human disease. In addition to classic nucleotide changes in the coding region of genes, positional and epigenetic effects gain increasing importance. This course discusses state-of-the-art research papers that will provide recent insights in new genetic mechanisms at the basis of human disease. Other topics include new molecular genetic analysis techniques, recent insights in the pathogenetic mechanisms and the therapeutic applications. Special attention is paid to the role of genetics in the development of new therapeutic strategies. Students will gain insight in diverse aspects of human genetics of the 21st century.

Contents

The following topics will be covered:
1. Epigenetics and imprinting
2. DNA methylation en histonacetylation
3. MicroRNA and other non protein coding RNAs
4. Disease mechanisms (haplo-insufficiency, dominant negative, gain-of-function, triplet repeat)
5. Long range regulation of transcription, conserved non-genic sequences
6. Non-mendelian inheritance/pedigree analysis
7. Complex inheritance
8. Mitochondrial genetics
9. Pharmacogenetics
10. Gene therapy
11. Preimplantation Genetic Testing

Initial competences

Having successfully completed the courses Human genetics, Molecular biology I, Molecular Biology II and Human pathogenesis from the bachelor program biomedical sciences, or having acquired the relevant ending objectives by other means.

Having completed succesfully the bachelor degree in biomedical sciences or having acquired the relevant ending objectives by other means.

Final competences

1 To gain new insights in the most actual developments of gene discovery, pathogenetic mechanisms and therapeutic strategies of human genetic conditions.
2 To be able to critically read and discuss scientific papers concerning gene discovery, pathogenic mechanisms and therapeutic strategies.
3 The student is able to express him/herself verbally in English, in such a way that the student can present and explain data of a state-of-the-art research manuscript.
4 The student is encouraged to ask questions (English) to peers during peer teaching and to the teaching professor during lectures.

Conditions for credit contract

Access to this course unit via a credit contract is determined after successful competences assessment

Conditions for exam contract

This course unit cannot be taken via an exam contract

Teaching methods

Lecture, Peer teaching

Extra information on the teaching methods

lectures and peer teaching

Study material

None

References

- state-of-the-art articles

Course content-related study coaching

Interactive support via questions and answers during lectures and by email/MS Teams.

Assessment moments

end-of-term assessment

Examination methods in case of periodic assessment during the first examination period

Written assessment

Examination methods in case of periodic assessment during the second examination period

Written assessment

Examination methods in case of permanent assessment

Peer and/or self assessment

Possibilities of retake in case of permanent assessment

not applicable

Extra information on the examination methods

Written assessment and peer assessment (microteaching). Students need to participate to both assessments.

Calculation of the examination mark

The written assessment accounts for 70% and the peer assessment (microteaching) for 30%.