Doctoral Program in Medical Genetics and Genomics

Mandatory subjects

Program Director: *Prof. Dr. Márta Széll, head of department, full professor*

Clinical Genetics I (Lead instructor: Prof. Dr. Márta Széll; 28 hours/6 credits, semester 2 or 4)

Clinical Genetics II (Lead instructor: Prof. Dr. Márta Széll; 28 hours/6 credits, semester 2 or 4)
Actualities in Human Genetics (or Current Developments in Human Genetics) (Lead instructor: Prof. Dr. Márta Széll; 28 hours/6 credits, semester 2 or 4)

Course description

Informing students on course requirements

Program: PhD full-time training
Course: **Clinical Genetics 1**
Course code: **IODI-MEDGE-01**
Academic year/Semester: 1st or 2nd year of the program, spring semester
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Type of course: lecture
Total number of hours in the subject: 28 Weekly hours of the course: 2
Credit vale of the course: 6
Type of examination: written five-step report
Preliminary requirements: none
Purpose of course:

The aim of the course is to provide students with a comprehensive, up-to-date and clinically focused knowledge of the basic principles of medical genetics and genomics, the main mechanisms of human inheritance, the genetic background of monogenic and complex diseases, and the clinical diagnostic use of modern, latest genetic and genomic technologies. The course also aims to enable students to clinically interpret genetic results, understand the basics of genetic counseling, and recognize the role of genetics and genomics in prevention, screening programs, therapeutic decision-making, and personalized medicine.

Outcome requirements of the course:

After successfully completing the course, the student will have comprehensive knowledge of the molecular background, clinical presentation and inheritance of monogenic, chromosomal and multifactorial diseases. The student is able to review the indications and limitations of modern genomic diagnostic methods - including next-generation sequencing, CNV analysis and epigenetic tests - and is able to interpret basic genetic and genomic test results. The student is familiar with the process of genetic counseling, communication, ethical and legal contexts, is aware of the domestic and international regulatory background of human genetic research, and recognizes the role of genomic results in clinical decision-making, risk assessment, prevention and the development of personalized therapeutic strategies.

Topics:

1. Genetics and medicine. The human genome. Genome programs, the post-genomic era, new technologies
2. Chromosomal Abnormalities Clinic
3. Clinical features of dominant and recessive inheritance , diseases
4. Epigenetics
5. Multifactorial inheritance, Genetics of complex diseases , importance of gene-environment interactions in the development of complex diseases
6. Genetics of human reproduction . Prenatal genetic screening and diagnostics
7. Pharmacogenetics, pharmacogenomics
8. Teratogenesis. Teratogens in clinical practice. Dysmorphology.
9. Gene therapy
10. Risk assessment, new technologies
11. Genetic counseling. Ethical and legal aspects. Genetic law, gene banks. Local specificities: European Union, US, Hungary
12. Genetics and genomics of malignant diseases .
13. The use of genomics results in diagnostics, therapy and prevention of human diseases

Supporting methods to achieve learning outcomes:

The learning outcomes are supported by frontal, presentation-based theoretical lectures, illustrative case studies, current clinical examples, illustrative genomic data and variant interpretation examples, and an evidence-based approach based on the latest international recommendations and guidelines (ACMG/AMP, ESHG). The course emphasizes the development of a practice-oriented approach, the development of critical literature reading, and independent scientific orientation, with

particular attention to the clinical interpretation and application of modern genomic technologies. The methodological elements highlighted during the education are interactive instructor-student discourse, guided literature selection, and the inclusion of current research examples, which promote a deeper understanding and integration of clinical genetic thinking.
Evaluation of the acquisition of expected learning outcomes: The assessment of the acquired material is carried out in the form of a written report, which contains integrative questions covering the entire subject area and assesses the students' theoretical knowledge, understanding of genetic and genomic relationships, and their clinical applicability. During the exam, we measure the students' ability to synthesize the concepts, methodologies, variant interpretation aspects, and clinical examples learned during the lectures, and to reflect the modern genomics diagnostic, prognostic and therapeutic aspects. The assessment is done on a five-point grading scale.
Mandatory reading list: 1. Éva Oláh: Clinical Genetics. Medicina Publishing. Year of publication: 2015. 2. Peter D Turnpenny: Emery,s Elements of Medical Genetics. Elsevier Health Sciences Publishing. Year of publication: 2021. https://shop.elsevier.com/books/emerys-elements-of-medical-genetics-and-genomics/turnpenny/978-0-7020-7966-5
Recommended reading list: 1. Andrew P Read, Dian Donnai: New Clinical Genetics. Scion Publisher. Publication year: 2020. https://www.amazon.com/Clinical-Genetics-fourth-Andrew-Read/dp/1911510703 2. Ronald Cohn, Stephen Scherer, Ada Hamosh: Thompson & Thompson Genetics and Genomics in Medicine. Elsevier Health Sciences Publisher. Publication year: 2023. https://www.libristo.hu/hu/konyv/thompson-thompson-genetics-and-genomics-in-medicine

Course description

Informing students on course requirements

Program: PhD full-time training, mandatory subject
Course: **Clinical Genetics 2**
Course code: **IODI-MEDGE-02**
Academic year/Semester: 1st or 2nd year of the program, spring semester
Educator and contact details (e-mail): Prof. Dr. Márta Széll, email: szell.marta@med.u-szeged.hu

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Type of course: lecture

Total number of hours in the subject: 28
Weekly hours of the course: 2

Credit vale of the course: 6

Type of examination: written five-step report

Preliminary requirements: none

Purpose of course:

The aim of the course is to provide students with a comprehensive, practical understanding of the methodological foundations, clinical applicability and limitations of modern molecular genetic and genomic studies. The course provides a systematic overview, from traditional cytogenetics to next-generation sequencing platforms and bioinformatics processing to long-read technologies and functional genomics approaches. Special emphasis is placed on real-life clinical examples, especially in the areas of oncogenetics, neurogenetics and genodermatosis. Students will be introduced to the critical steps of the diagnostic workflow: sample handling – data acquisition – bioinformatics – interpretation – clinical report. The course supports a research and translational mindset and promotes competent participation in multidisciplinary collaborations. Upon completion of the course, students will be able to apply genetic results in an evidence-based, clinically meaningful manner.

Outcome requirements of the course:

Upon completion of the course, the student will have a comprehensive understanding of the indications, advantages and limitations of modern cytogenetic, molecular genetic, next-generation sequencing and epigenetic testing methods. The student will be able to interpret critical steps in the diagnostic and research workflow, including preanalytical factors, library preparation, sequencing platforms, bioinformatics evaluation, quality control, variant annotation and clinical interpretation. The student will recognize technical, bioinformatics, statistical and clinical interpretation limitations and apply critical considerations when evaluating results. The student will be able to interpret variant annotation outputs at a basic level and understand the hierarchy of evidence levels (e.g. ACMG/AMP guidelines, population frequencies, relevance of functional data). Completion of the course will enable transdisciplinary communication and the formulation of clinically relevant, well-defined questions when developing a genetic testing strategy.

Topics:

14. Traditional cytogenetic testing methods and their applications (karyotyping , FISH)
15. MLPA and array -CGH tests
16. Traditional PCR methods and sequencing (RT-PCR, qPCR, digital PCR, capillary sequencing)
17. New generation sequencing (NGS) theory and laboratory methodologies, sample preparation
18. What's new in long-read whole genome sequencing?
19. Functional testing methods: NIPTs
20. Chromosomal abnormalities (numerical and structural abnormalities) – with cases
21. Oncohaematological methods
22. Bioinformatics methods
23. Array CGH, optical genome mapping
24. Epigenetic studies

Supporting methods to achieve learning outcomes:

The learning outcomes are supported by frontal lectures, case-based illustrations, methodological demonstrations, integration of current literature examples, as well as guided discussions. The course emphasizes interactive questioning that encourages critical interpretation and the strengthening of decision-support thinking through real clinical and research examples.

Evaluation of the acquisition of expected learning outcomes:

The evaluation of the expected learning outcomes takes the form of a written exam, which aims to assess methodological knowledge, application areas, and interpretation and critical analysis skills. The exam tasks measure the understanding of the key steps of the genomic diagnostic workflow, the overview of the indications and limitations of the methodologies, and the correct application of basic concepts related to variant interpretation. The students should give a short (10 minutes + 5 minutes discussion) student presentation once based on a paper related to a current methods, which does not qualify as a grade-affecting exam section, but is a condition for admission to the exam. The basis of the assessment is the written report.

Mandatory reading list:

hand-out

Recommended reading list:

Methodological scientific publications downloadable through targeted PubMed search.

Course description

Informing students on course requirements

Program: PhD full-time training
Course: **Actualities in human genetics**
Course code: **IODI-MEDGE-03**
Academic year/Semester: 1st or 2nd year of the program, spring semester
Educator and contact details (e-mail) Prof. Dr. Márta Széll, email: szell.marta@med.u-szeged.hu Dr. Nikoletta Nagy, email: nagy.nikoletta@med.u-szeged.hu Dr. Zsuzsanna László, email: laszlo.zsuzsanna@med.u-szeged.hu Dr. Tibor Kalmár, email: kalmar.tibor@med.u-szeged.hu Dr. Zoltán Maróti, email: maroti.zoltan@med.u-szeged.hu Dr. Krisztina Búzás, email: buzas.krisztina@med.u-szeged.hu Dr. Sára Tóth, email: toth.sara@med.u-szeged.hu Prof. Dr. Ernő Duda, email: duda.erno@med.u-szeged.hu Dr. Judit Danis, email: danis.judit@med.u-szeged.hu Dr. Lőrinc Pongor, email: lorinc.pongor@hcemm.eu
Type of course: lecture
Total number of hours in the subject: 28 Weekly hours of the course: 2
Credit vale of the course: 6
Type of examination: written five-step report
Preliminary requirements: none
Purpose of course: The aim of the course is to provide students with a comprehensive and critical view of the latest scientific findings and trends in human genomics. The course introduces the main research programs of the post-genomic era and their clinical translational potential. Special emphasis is placed on the latest mechanistic knowledge about non-coding RNAs and their functional role in human diseases. Students gain insight into the evolutionary genetic background of human diseases and their interpretational characteristics. The course also aims to develop a structured, evidence-based analysis of ethical aspects, especially in the context of modern genomic diagnostic practice. The subject promotes the enhancement of research-interpretation competence, the strengthening of independent, critical literature use skills, and the development of autonomy in scientific decision support.

Outcome requirements of the course:

The student knows the objectives and translational significance of the main genome programs of the post-genomic era. Recognizes and can justify the ethical challenges related to clinical genetic decision-making. Is able to compare the evolutionary genetic background of different human diseases, along with their interpretation characteristics. Is able to build structured, evidence-based reasoning in modern genomic diagnostic situations. Presents and applies critical evaluation aspects of the significance and limitations of modern genome data analysis. Presents in detail the role of non-coding RNAs in human disease processes, at the mechanism level. Independently interprets scientific publications on human genomics, with critical literature interpretation skills. Integrates the course content autonomously into his/her own doctoral research topic.

Topics:

1. The importance of the Human Genome Project in medical sciences (4 hours)
2. Genome programs of the post-genomic era and their significance in medical sciences (4 hours)
3. Non-coding RNAs and their role in human diseases – miRNAs (4 hours)
4. Non-coding RNAs and their role in human diseases – long non-coding RNAs (4 hours)
5. Evolutionary genetic aspects of human diseases (4 hours)
6. Ethical challenges of clinical genetics in the 21st century (4 hours)
7. Student presentations on course topics (4 hours)

Supporting methods to achieve learning outcomes:

The course primarily uses frontal lectures, which aim to present the latest human genomics results in a structured and systematic manner. Students participate in guided article discussions in seminar-like sessions, which develop critical literature reading and argumentation skills. Student presentations will also be held during the course, which serve to develop research result communication and scientific argumentation. The emphasis of the methodology is on the practical application of evidence-based thinking and the comparative evaluation of current genomic research directions. The educational strategy of the course specifically supports the development of autonomous research competence and integration into the doctoral research topic.

Evaluation of the acquisition of expected learning outcomes:

The condition for passing the subject is to be certified to attend at least 70% of the course sessions. During the semester, students give a short (10 minutes + 5 minutes discussion) student presentation once based on a paper related to a current genomics topic, which does not qualify as a grade-affecting exam section, but is a condition for admission to the exam. The end-of-semester assessment takes the form of a written colloquium, which covers key areas of the subject content (post-genomic programs, ncRNAs, evolutionary genetics, ethical aspects). The exam is assessed using a five-level grading system, which is determined based on the scores

obtained. A failed exam can be repeated according to institutional rules. Absence on exam days must be officially certified.
Mandatory reading list: hand-out
Recommended reading list: Scientific publications downloadable with targeted PubMed search.