REQUIREMENTS

Semmelweis University, Faculty of Medicine
Name(s) of the Institute(s) teaching the subject: Institute of Genomic Medicine and Rare Disorders

Name of the subject: Clinical genetics

Credits: 2

Total number of hours: **lectures:** 0 **practices:** 20 **seminars:** 8

Type of the course (mandatory/elective): mandatory

Academic year: 2022/23

Code of the course[1]: AOKGRI764_1A

Course director (tutor): Prof. Dr. Mária Judit Molnár

Contact details: SE Genomikai Medicina és Ritka Betegségek Intézete

Institute of Genomic Medicine and Rare Disorders; Semmelweis University, Faculty of Medicine

Position: Lecturer and Chairman of the Department, full professor

Date of habilitation and reference number: 38/ 2006 Debreceni Egyetem

Aim of the subject and its place in the curriculum:
The Clinical Genetics course introduces medical students to the clinical significance of human genetic variations, covering the principles of diagnosis and treatment of rare diseases, mainly through the presentation of cases, clinical situations and demonstrating correct communication. In addition to diagnosis of monogenic diseases and chromosomal abnormalities, genetic tests are now used to predict susceptibility to certain diseases or the efficacy and side effects of certain therapeutic options (pharmacogenomics) or delineate disease prognosis. The role of the physician in the management of hereditary diseases and the practice of good decision making will also be addressed. The discipline is one of the most rapidly developing field and it is presumably going to be an important component of the future medicine.

Location of the course (lecture hall, practice room, etc.):
seminars - Elméleti Orvostudományi Kar – EOK , seminar room 0.806 (1094 Budapest, Tűzoltó Str. 37-47.)
seminars - Semmelweis University Central Patient-Care Unit (Korányi Tömb, Központi Betegellátó Épület) 1082 Budapest Üllői Str. 78/B Building „A" (Radiology Wing) 4th floor
Neurogenetic Clinic - Semmelweis University Central Patient-Care Unit (Korányi Tömb, Központi Betegellátó Épület) 1082 Budapest Üllői Str. 78/B Building „B" (Main building) 1st floor
In-patient unit - Centre for Rare Disorders; Neurology Clinic – Neurology wing, 1st floor; 1083 Budapest Balassa János Str. 6.

Competencies gained upon the successful completion of the subject:

Students meet the typical clinical scenarios requiring genetic-genomic knowledge for diagnostic, predictive, preventive and pharmacogenomic questions, as well as they are introduced into most common diagnostic, therapeutic decision-making, management and ethical dilemmas of the field.

Students gain insight into practice how to elicit, document, and act on relevant family history pertinent to the patient's clinical status, how to choose the right genetic or genomic testing to guide patient management and how to use genomic information to make treatment decisions.

Through problems represented by several real and synthetic/standardized clinical cases and through getting involved into the patient flow of in- and outpatient units, they will learn

- how to gather information from appropriate information resources,
- how to employ artificial intelligence-based decision-making systems for phenotype pattern recognition an a problem-based manner.
- how to communicate with the patients and families with hereditary diseases by playing the role of a counselee (genetic counselling), and
- how to work together as a team if they are encountered with real-world diagnostic challenges.

Prerequisite(s) for admission to the subject:

Successful completion of Pharmacology II, Genetics and Genomics, Pathology II.

Minimum and maximum number of students registering for the course:

min.5 max. 22

Student selection method in case of oversubscription:

along the constraints of the block education

How to register for the course:

NEPTUN

Detailed thematic of the course[2]:

Lectures

0

Practices

Monday	8:00	Clinical genetics in medicine seminar
	10:00	Web-based search methods in clinical genetics workshop
	12:30	Bedside practice: inpatient department small group work
	14:30	Case presentation part 1small group work
Tuesday	8:00	The significance and diagnostic potential of congenital genetic disorders seminar
	10:00	Developing genetic diagnostic skills workshop
	12:30	Bedside practice: neurogenetic clinic small group work
	14:30	Case presentation part 2small group work
Wednesday	8:00	Genetic disorders of public health significance seminar
	10:00	Genetic counselling workshop, simulated counselling situation, role play
	12:30	Management of patients with a rare disease workshop
	14:30	Team competition decision-making simulation, team-based competition
Friday	8:00	Personalised medicine seminar
	10:00	Ethical problems, pre-exam consultation workshop, consultation
	12:30	Exam written, single choice test

Potential overlap(s) with other subjects:

preclinical module: Genetics and Genomics

Clinical module: pediatrics, neurology, cardiology, oncology, obstetrics-gynaecology, internal medicine, pharmacology

Special training activities required[3]:

use a digital workbook, make a presentation, perform IT tasks

Policy regarding the attendance and making up absences:

In accordance with the study and examination regulations, attendance is compulsory for 75% of the sessions, which also applies specifically to the bedside classes in the in- and outpatient units, genetic counselling role play and team competition classes.

Means of assessing the students' progress during the semester[4]: During the short teaching period available, there is no interim formal assessment. However, the interactive nature of the exercises and consultations will allow students' knowledge and use of the information available to them to be monitored.
Requirement for acknowledging the semester (signature): At least 75% attendance in the sessions, attendance is checked at every classes.
Type of the examination: written, online
Exam requirements[5]: Problem-oriented written test questions evenly covering the topics detailed above
Type and method of grading[6]: 61% of the points is needed to pass, with equal intervals above this
How to register for the exam: NEPTUN
Opportunities to retake the exam: according to Regulations of the university in force
Literature, i.e. printed, electronic and online notes, textbooks, tutorials (URL for online material): 1- E-learning material (szerk. Dr. Molnár Mária Judit) (2018) 2- Turpenny P, Ellard S: Emery's Elements of Medical Genetics (2012)
Signature of the tutor:
Signature(s) of the head(s) of the Institute(s):
Date:

Credit Transfer Committee's opinion:
Comment of the Dean's Office:
Signature of the Dean:

[1] Dékáni Hivatal tölti ki, jóváhagyást követően.
[2] Az elméleti és gyakorlati oktatást órákra (hetekre) lebontva, sorszámozva külön-külön kell megadni, az előadók és a gyakorlati oktatók nevének feltüntetésével. Mellékletben nem csatolható!
[3] Pl. terepgyakorlat, kórlapelemzés, felmérés készítése stb.
[4] Pl. házi feladat, beszámoló, zárthelyi stb. témaköre és időpontja, pótlásuk és javításuk lehetősége.
[5] Elméleti vizsga esetén kérjük a tételsor megadását, gyakorlati vizsga esetén a vizsgáztatás témakörét és módját.
[6] Az elméleti és gyakorlati vizsga beszámításának módja. Az évközi számonkérések eredményeink beszámítási módja.