

REQUIREMENTS

Semmelweis University, Faculty of General Medicine – single, long-cycle medical training programme

Name of the host institution (and any contributing institutions):

Department of Endocrinology, Department of Internal Medicine and Oncology

Name of the subject:

in English: Genetics of endocrine diseases

in German: Genetik der endokrinen Erkrankungen

Credit value: 2

Semester: 2.

(as defined in the curriculum)

Total number of classes per week:	2	lectures:	X	practical lessons:		seminars:	
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Type of subject: compulsory optional elective

(PLEASE UNDERLINE AS APPLICABLE)

Academic year: 2023/2024, every year

Language of instruction, for optional or elective subjects: English

Course code: AOS EDT869_1A

(In the case of a new subject, this cell is filled in by the Dean's Office, following approval)

Course coordinator: Prof. Dr. Peter Igaz

Place of work, phone number: Department of Endocrinology, Department of Internal Medicine and Oncology, Tel: 2660816

Position: Head of the Department of Endocrinology

Date and number of habilitation: 2012, 333

Objectives of the course and its place in the medical curriculum:

There are several very interesting, rare diseases in the field of endocrinology that are caused by genetic alterations. By getting acquainted with the pathomechanism and clinical picture of these diseases, the students can refresh their genetic and endocrinological knowledge, moreover can get novel knowledge in genetics and in endocrinology. The course presents among others the features of hereditary endocrine tumor syndromes, enzyme defects and hormone resistance syndromes.

Place of instruction (address of lecture hall or seminar room etc.):

Teaching facilities of the Department of Internal Medicine and Oncology, 1083 Budapest, Korányi S. str. 2/a – Lecture hall

Competencies acquired through the completion of the course:

By getting acquainted with the pathomechanism and clinical picture of these diseases, the students can refresh their genetic and endocrinological knowledge, moreover can get novel knowledge in genetics and in endocrinology. The course presents among others the features of hereditary endocrine tumor syndromes, enzyme defects and hormone resistance syndromes.

Prerequisites for course registration and completion:

Internal Medicine I., Genetics and genomics

Conditions for concurrent course registration and permission thereof in the case of a multi-semester subject: -**Student headcount conditions for starting the course (minimum, maximum) and method of student selection:**

min. 5., max. 50 students

Detailed course description:

(Theoretical and practical instruction must be broken down into lessons (weeks), numbered separately. Please provide the names of lecturers in both types of lessons, indicating guest lecturers. This information is not to be attached separately. CVs of guest lecturers, however, must be attached.)

1. Overview of endocrine diseases with genetic background. Main disease groups and genetic mechanisms. (Dr. Igaz Péter, Department of Endocrinology, Department of Internal Medicine and Oncology)
2. Multiple Endocrine neoplasia syndromes. (Dr. Igaz Péter, Department of Endocrinology, Department of Internal Medicine and Oncology)
3. Genetics of pheochromocytoma (Dr. Patócs Attila, Department of Laboratory Medicine)
4. Genetics of hyperaldosteronism, hypercortisolism and adrenocortical cancer (Dr. Igaz Péter, Department of Endocrinology, Department of Internal Medicine and Oncology)
5. Genetics of pituitary tumors and hypopituitarism (Dr Butz Henriett, Department of Laboratory Medicine)
6. Relevance of Ca-sensor mutations in endocrinology (Dr. Tőke Judit, Department of Endocrinology, Department of Internal Medicine and Oncology)
7. Genetics of thyroid tumors (Dr. Lakatos Péter, Department of Endocrinology, Department of Internal Medicine and Oncology)
8. Hormone resistance syndromes (thyroid hormone resistance, pseudohypoparathyroidism, glucocorticoid resistance, androgen insensitivity) (Dr. Igaz Péter, Department of Endocrinology, Department of Internal Medicine and Oncology)
9. Genetics of congenital adrenal hyperplasia (Dr. Patócs Attila, Department of Laboratory Medicine)
10. Genetics of type 1 and type 2 diabetes mellitus (Dr. Lukács Krisztina, Department of Internal Medicine and Hematology)
11. Clinical picture and genetics of monogenic diabetes mellitus (MODY) (Dr. Kocsis Győző, Department of Internal Medicine and Oncology)
12. Chromosome alterations with endocrine relevance (Turner-syndrome, Klinefelter-syndrome, Prader-Willi-syndrome) (Dr. Haltrich Irén, 2nd Dept. of Pediatrics)
13. Genetics of rare metabolic diseases (Dr. Reismann Péter, Department of Endocrinology, Department of Internal Medicine and Oncology)
14. Test exam

Related subjects due to interdisciplinary fields (both compulsory and elective) and potential overlaps between subjects:

The topics of some lectures overlap with some topics of endocrinology in Internal Medicine I, and the elective course Clinical Endocrinology, but in this optional course, the focus is on the genetics of endocrine diseases, therefore the topics are presented from different aspects and focus.

Attendance requirements; conditions under which students can make up for absences and the

method of absence justification:

3 absences permitted

Form of assessment in the study period:

(including the number, topics and scheduling of oral and written tests, their share in the overall evaluation, make-up tests and improvement tests)

Number and type of assignments for individual work and the deadline for submission:

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Requirements to obtain the teacher's signature:

3 absences permitted

Type of assessment (*comprehensive examination, end-term examination, term-grade, term-grade on a three-grade rating scale, no examination*):

Multiple choice test questions, min. 60 % right answers are required for a successful exam.

Examination requirements:

(*list of examination topics, subject areas of tests / examinations, lists of mandatory parameters, figures, concepts and calculations, practical skills*)

The topics are defined by the lectures, whose materials will be presented following the lectures on Moodle and on the homepage of the Department of Internal Medicine.

Possibilities for exam retake: oral exam (with the course leader Prof. Peter Igaz)

Method and type of grading:

(*Share of theoretical and practical examinations in the overall evaluation. Inclusion of the results of the end-of-term assessment. Possibilities of and conditions for offered grades.*)

Written test.

Multiple choice questions, min. 60 % right answers for a successful exam.

60-70 % - 2

70-80 % - 3

80-90 % - 4

90-100 % - 5

List of course books, textbooks, study aids and literature facilitating the acquisition of knowledge to complete the course and included in the assessment, precisely indicating which requirement each item is related to (e.g., topic by topic) as well as a list of important technical and other applicable study aids:

- Lectures
- Igaz P & Patócs A: Genetics of Endocrine Diseases and Syndromes, Springer, 2019
<https://www.springer.com/gp/book/9783030259044>

Signature of habilitated instructor (course coordinator) announcing the course:

Signature of the director of the host institution:



Date of submission:

20.07.2023.