Study programme: REPRODUCTIVE BIOLOGY

Course title: Preimplantation genetic diagnosis

Teacher: Mihajla Djan

Course status: elective

ECTS: 4

Requirements: -

Course objectives

The aim of this course is to introduce students to the principles of preimplantation genetic diagnosis (PGD) and procedures used in preimplantation genetic diagnostics for hereditary diseases of different etiologies.

Learning outcomes

After successfully realized pre-exam and exam obligations student can:
- understand the procedures used in the preimplantation genetic diagnosis
- explain the genetic basis of hereditary disorders
- define the principles of diagnosis of chromosomal aberrations, monogenic diseases, sex-linked diseases
- understand the basics of prenatal diagnosis
- use internet resources and literature in the field of preimplantation genetic diagnostics

Syllabus

Theoretical instruction

The history and development of preimplantation genetic diagnosis (PGD). The procedures used in PGD. The genetic basis of hereditary diseases. Preimplantation Genetics. PGD of chromosomal aberrations. PGD of monogenic diseases. PGD of mitochondrial diseases. PGD in clinical cases of infertility. PGD of sex-linked diseases and non-medical sex selection. Preimplantation genetic screening (PGS). Basics of prenatal diagnosis.

Research activities

Karyotyping and diagnosis of chromosomal aberrations - virtual cases. Use of internet resources of genetic basis of hereditary diseases - OMIM. Use of internet sources on the procedures and tests in preimplantation genetic diagnostics. Discussions about the latest scientific information in the field of preimplantation genetic diagnosis - journal club.

Literature

1. Harper JC. Preimplantation Genetic Diagnosis. Second Edition. Cambridge University Press. 2009.
2. Strachan T., Read AP. Human Molecular Genetics 3. Garland Publishing, New York, USA. 2004.
3. Turnpenny P., Ellard S., Emery's elements of medical genetics, 12th Ed., Elsevier, Churchill Livingstone, 2005.
4. Review papers in the field of preimplantation genetic diagnosis.

Weekly teaching load	**Lectures:** 2	**Practical lectures:** 0+0+1

Teaching methods

Lectures, practical lectures, seminar, tuition

Evaluation of knowledge (maximum score 100)

Pre-exam obligations	points	**Final exam**	points
Student engagement in lectures		Final test	
Student engagement in practical lectures		Oral exam	60
seminars	40		