GENETIC SYNDROMES THAT DEFINE NEURO-DEVELOPMENTAL DISEASES

(1) GENERAL

SCHOOL	HEALTH SCIENCES		
ACADEMIC UNIT	SPEECH LANGUAGE THERAPY		
LEVEL OF STUDIES	Undergraduate Program (Level 6)		
COURSE CODE	slt – 610	**SEMESTER**	**6th**
COURSE TITLE	Genetic syndromes that define neuro-developmental diseases		
INDEPENDENT TEACHING ACTIVITIES *if credits are awarded for separate components of the course, e.g. lectures, laboratory exercises, etc. If the credits are awarded for the whole of the course, give the weekly teaching hours and the total credits*		**WEEKLY TEACHING HOURS**	**CREDITS**
lectures		3	4
COURSE TYPE *general background, special background, specialised general knowledge, skills development*	Specialised General Knowledge		
PREREQUISITE COURSES:	No		
LANGUAGE OF INSTRUCTION and EXAMINATIONS:	Greek and English		
IS THE COURSE OFFERED TO ERASMUS STUDENTS	YES		
COURSE WEBSITE (URL)	https://slt.uoi.gr/		

(2) LEARNING OUTCOMES

Learning outcomes

The course learning outcomes, specific knowledge, skills and competences of an appropriate level, which the students will acquire with the successful completion of the course are described.

Consult Appendix A

- *Description of the level of learning outcomes for each qualifications cycle, according to the Qualifications Framework of the European Higher Education Area*
- *Descriptors for Levels 6, 7 & 8 of the European Qualifications Framework for Lifelong Learning and Appendix B*
- *Guidelines for writing Learning Outcomes*

The aim of the course is for students to know the basic principles of genetics as well as genetic syndromes that define various neurodevelopmental diseases.

Upon successful completion of the program the student will be able to:

- Adequately know the basic principles of genetics (Levels 1 & 2: Knowledge & Understanding)
- Recognize deformed characteristics that would indicate a genetic disease (Levels 1, 2 & 3: Knowledge, skill & ability)
- understand the techniques of genetics and genetic analysis (karyotype, Next Generation Sequencing, Whole Exome Sequencing, Whole Genome Sequencing, Gene panels). (Levels 1 & 2: Knowledge & Understanding)
- be trained in the genetic syndromes that define a variety of neurodevelopmental diseases (Levels 1, 2, 3 & 5: Knowledge, skill, ability & Composition)

General Competences

Taking into consideration the general competences that the degree-holder must acquire (as these appear in the Diploma Supplement and appear below), at which of the following does the course aim?

Search for, analysis and synthesis of data and information, with the use of the necessary technology
Adapting to new situations
Decision-making
Working independently
Team work
Project planning and management
Respect for difference and multiculturalism
Respect for the natural environment
Showing social, professional and ethical responsibility and sensitivity to gender issues
Criticism and self-criticism

Working in an international environment	*Production of free, creative and inductive thinking*
Working in an interdisciplinary environment	*……*
Production of new research ideas	*Others…*
	…….

- *Search for, analysis and synthesis of data and information, with the use of the necessary technology*
- *Decision-making*
- *Working independently*
- *Team work*
- *Working in an international environment*
- *Working in an interdisciplinary environment*
- *Production of new research ideas*
- *Project planning and management*
- *Respect for difference and multiculturalism*
- *Showing social, professional and ethical responsibility and sensitivity to gender issues*
- *Criticism and self-criticism*
- *Production of free, creative and inductive thinking*
- *Research work*
- *Writing of research paper*
- *Collaboration with relevant specialties*

(3) SYLLABUS

1. Basic principles of genetics
2. Human genome and the chromosomal basis of heredity
3. Neurogenetics
4. Recognition of deformed characteristics
5. Modern methods of laboratory analysis A
6. Modern methods of laboratory analysis A
7. Concept of mutations
8. Ethical issues and genetics
9. Genetic syndromes that define neurodevelopmental diseases A
10. Genetic syndromes that define neurodevelopmental diseases B
11. Prenatal diagnosis
12. Genetic Councelling
13. Treatment of genetic diseases

(4) TEACHING and LEARNING METHODS - EVALUATION

DELIVERY *Face-to-face, Distance learning, etc.*	Face-to-face
USE OF INFORMATION AND COMMUNICATIONS TECHNOLOGY *Use of ICT in teaching, laboratory education, communication with students*	Use of audio-visual methods (e.g. PowerPoint and Video presentations) Support the learning process through the e-class platform.
TEACHING METHODS *The manner and methods of teaching are described in detail.* *Lectures, seminars, laboratory practice, fieldwork, study and analysis of bibliography, tutorials, placements, clinical practice, art workshop, interactive teaching, educational visits, project, essay writing, artistic creativity, etc.* *The student's study hours for each learning activity are given as well as the hours of non-directed study according to the principles of the ECTS*	

Activity	***Semester workload***
Lectures	39
Study and analysis of bibliography	5
Essay writing	10
Research Work	5
Personal Study/Evaluation	41
Course total	100

STUDENT PERFORMANCE EVALUATION *Description of the evaluation procedure* *Language of evaluation, methods of evaluation, summative or conclusive, multiple choice questionnaires, short-answer questions, open-ended questions, problem solving, written work,*	**I.** Written final exam (80%): - Multiple choice test - Short answer questions **II.** Individual and/or Teamwork Written Essay (20%)

essay/report, oral examination, public presentation, laboratory work, clinical examination of patient, art interpretation, other *Specifically-defined evaluation criteria are given, and if and where they are accessible to students.*	(with Pass, Merit and Distinction criterion accessible by students) The final exams will be offered in Greek & English

(5) ATTACHED BIBLIOGRAPHY

-Suggested Bibliography:

- *Klug WS, Cummings MR, Spencer CA, Palla MA (2019) Βασικές Αρχές Γενετικής, Εκδόσεις Μπάσδρα Ι. I***SBN:** 978-618-5135-18-8 *[Προτεινόμενη]*
- *Thomson and Thomson (2011). Ιατρική Γενετική, Ιατρικές Εκδόσεις Πασχαλίδης, Αθήνα*

-Relevant Scientific Journals:

- Neurogenetics, https://www.springer.com/journal/10048
- Genetics, https://www.genetics.org/
- Nature genetics, https://www.nature.com/ng/