# **Human genetics (IIInd, Vrd SEMESTER)**

## Credit value (ECTS) 9

## **Course category**

Domain (Imposed)

### **Course holder:**

Assoc. Prof. PhD. Silvica PĂDUREANU

## Discipline objectives (course and practical works)

The course aims to make students acquire knowledge on the interpretation of human phenotype based on the interaction between genotype and environment. It is also aims to understand the way in which the individuality (unique) for each human individual, the manner in which the biological polymorphism in the human population, the relationship between heredity and disease.

Practical work aims to familiarize students with the compilation and interpretation of the family tree, the mode of transmission of monogenic hereditary disease in the human population, with the manner of determining paternity and filiation based on blood groups.

## **Contents (syllabus)**

Course (chapters/subchapters)		
Introduction to human genetics		
Man, heredity and environment		
Human chromosomes		
Genetics embryonal		
Cellular basis of heredity and human variability		
The human reproduction		
Monogenic human transmission characters		
Polygenic character transmission to humans		
Elements of medical genetics		
Elements of Cancer Genetics		

### **Practical works**

Segregation in the pedigree: how to prepare a family tree.

Factors that alters the equilibrium of Hardy-Weinberg law in human populations.

How applies Hardy-Weinberg law at human monogenic diseases: autosomal dominant disease, autosomal recessive disease, recessive disease linked to chromosome X.

Highlighting sexual chromatin.

Transmission characters monogenic hereditary determinism normal in humans.

Final colloquium of knowledge evaluation

## **Bibliography**

- 1. Covic M., Ștefănescu D., Sandovici I., 2011 Genetica medicală, Ed. Polirom Iași
- 2. Gavrilă L., 2003 Genomica, Editura Enciclopedica, Bucuresti
- 3. Gavrilă L., 2004 Principii de ereditate umană, Editura BIC ALL, București
- 4. Gavrilă L., 2004 Genomul uman, Editura BIC ALL, București
- 5. Gorduza E.V., Stoica O.F., 2003 Elemente de genetica umană, Ed. Timpul, Iași
- 6. Ierima A., 2005 Genetica umană., Ed. Zigotto, Galați
- 7. Marc J., 2004 Génétique médicale: formelle, chromosomique, moléculaire, clinique, Masson, Pari
- 8. Maximilian C., 1986 Genetica medicală, Ed. Medicală, București
- 9. Munteanu I., 2006 Tratat de Obstetrică și Ginecologie, Academia Română, București
- 10. **Pădureanu Silvica, 2014** Genetica umană Note de curs, U.S.A.M.V., Iași
- 11. Serre Jean-Louis, 2004 Génétique medicale: rappels des cours, exercices et problèmes corrigés, Dunod, Paris

### **Evaluation**

Evaluation form	Evaluation Methods	Percentage of the final grade
Exam	Oral examination	60%
	Oral assessment during the semester, verification tests and final laboratory colloquium.	40%

#### Contact

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