

Human genetics (IIIInd, Vrd SEMESTER)

Credit value (ECTS) 7

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 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: classification, characteristics.
Genetics of embryonic development.
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, București
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. Pădureanu Silvica, 2014 – *Genetica umană – Note de curs*, U.S.A.M.V., Iași
7. Serre Jean-Louis, 2004 - *Génétique médicale: rappels des cours, exercices et problèmes corrigés*, Dunod, Paris

Evaluation

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

Contact

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