| Subject | BASICS IN HUMAN GENETICS | | | |
|------------------------|--|--|--|--|
| Study programme | Three-year expert studies for graduated speech therapists | | | |
| Code | CL 124 | | | |
| Study year | First (I) | | | |
| Semester | Second (II) | | | |
| Lessons in total | 60 | | | |
| Type of subject | Mandatory | | | |
| Requirements | None | | | |
| Executor | Cathedra of pediatrics, Cathedra of immunology and Cathedra of gynecology | | | |
| Responsible | Prof. d-r Mirjana Kocova | | | |
| professor | , | | | |
| Address | University pediatric clinic, Vodnjanska no.17, 1000 Skopje, tel. +00389 2 3111713; fax +389 2 323 224 email: mkocova@medf.ukim.edu.mk | | | |
| Key words | Studies for speech therapists, basic subject, basic in human genetics | | | |
| Learning objectives | Training the students about the basic genetic principles that influence medical practice Training the students regarding basic principles of cytogenetics, molecular genetics, biochemical genetics, population genetics, reproductive genetics and genetics in forensic medicine Educating the students on basic principles in communication with families with genetic disorders and malformations | | | |
| | Training the students about basic ethical principles in genetics | | | |
| Short contents | Theoretical education (30 lessons) • Basics in human genetics • Genetic disorders in medicine | | | |
| Short contents | Practical education (30 lessons) | | | |
| | Methods of genetic analysis Methods of genetic diagnosis, syndrome and malformation recognition, screening, genetic counseling | | | |
| Organisation | Theoretical training: 30 lectures | | | |
| | Practical training: 30 lectures | | | |
| Learning methods | Working in small groups, practicals, seminar work | | | |
| Provided study results | Knowledge and understanding: DNA as a genetic material. DNA replication. Genetic control of proteins. Genetic expression. Recombinant cloning of DNA. Mendel genetics. Gene mapping in prokaryotes and eukaryotes. Non-Mendelian inheritance. Regulation of gene expression. Mutations and polymorphisms. Clinical cytogenetics. Molecular and biochemical essentials of genetic diseases. Treatment of genetic diseases. Genetics of diseases with monogenetic and complex type of inheritance. Genetics of immune system and cancer genetics. Prenatal diagnosis. Genetic counseling, ethical aspects. | | | |
| | Key skills: Molecular genetics; Cytogenetics; Immunogenetics; Biochemical genetics; Communication with families; Taking data and genetic information; Methods for prenatal diagnosis. | | | |
| | Student is obliged to follow all planned activities, including participation | | | |

| | in continuous checking of kr | nowledge to receive | the signature | |
|--|--|---------------------|---------------|--|
| | Scorring of the student's activities: | | | |
| | Activity | Scoring | | |
| | , | Min. | Max. | |
| Specific recommendation for training | Theoretical education* | 4 | 6 | |
| | Practical education** | 18 | 30 | |
| | Seminars | 0 | 4 | |
| | Continuous checks -2 | 12 | 18 | |
| | Final exam | 26 | 42 | |
| | *Attendance in theoretical tra | 60 | 100 | |
| | **attendance in practicals: (each practical - 1 point) (30 practicals) Attendance 0,25 points Solving the practicals - 0.75 points Conditional criteria for knowledge verification: 1. For entering the final exam, student should acquire minimum points (60%) from the two continuous checks 2. If the student didn't reach minimum score from the two continuous checks, he approaches for the complete exam. | | | |
| Checking the knowledge | Continuous checks of knowledge-2 (in writing) 1. Basics in human genetics (30 questions) 2. Main genetic disorders (30 questions) 6-9 points Final exam* writing test+ practical exam + oral exam 1. Basics of human genetics and genetic disorders (40 questions that were not checked previously) 8-12 points 2. Practical exam - 1 practical from basics in human genetics and 1 practical in main genetic disorders 12-20 points 3. Analytical skills (integrating of genetic mechanisms) 6-10 points Complete exam* is a combination of results gained from continuous checks and final exam (writing test+ practical exam + oral exam) Assessment of whole exam is estimated according the table of received grades, based on the sum of points gained from all activities, including continuous checks and points from every part or final exam or complete exam. | | | |
| Teaching aids | Authorized lectures in human genetics-published - Prof. d-r M. Kocova and all Genetics 2006. Prof d-r M. Spiroski (compact disc) Practicum for human genetics 1 Prof.d-r M. Spiroski and all Practicum for human genetics 2 Prof.d-r M. Kocova and all Molecular diagnostics, Prof G. Efremov and Clinical Genetics in: Nikodievich B: Advanced diagnostics and therapy in medicine, Skopje, 2000 | | | |