| 1.  | Subject  | HUMAN GENETICS  |          |   |                       |                               |  |  |  |  |
|---|--|---|----------|---|-----------------------|-------------------------------|--|--|--|--|
| 2.  | Code   | MLD - 216   |          |   |                       |                               |  |  |  |  |
| 3.  | Study program:   | Three-year professional studies of medical laboratory diagnostics   |          |   |                       |                               |  |  |  |  |
| 4.  | Conducted by   | UKIM – Medical Faculty  |          |   |                       |                               |  |  |  |  |
| ~   |  | Department of Human Genetics  |          |   |                       |                               |  |  |  |  |
| 5.  | Degree of  | First cycle   |          |   |                       |                               |  |  |  |  |
|   | education (first or second cycle)  |   |          |   |                       |                               |  |  |  |  |
| 6.  | Academic   | II/III 7.   | Credits  |   | 2                     |                               |  |  |  |  |
|   | year/semester  |   |          |   |                       |                               |  |  |  |  |
| 8.  | Professor  | Prof. d-r Aleksandar Petlichkovski  |          |   |                       |                               |  |  |  |  |
|   |  | The lessons are held by all the members of the Department   |          |   |                       |                               |  |  |  |  |
| 9.  | Prerequisite   | None  |          |   |                       |                               |  |  |  |  |
| 10.   | Output       Goals       The students will be able to:         -       Define the basic terms of the human genetics field  |   |          |   |                       |                               |  |  |  |  |
|   |  |   |          |   |                       |                               |  |  |  |  |
|   |  |   |          |   | non-Mendel's laws     |                               |  |  |  |  |
|   |  |   |          |   |                       | netic diseases, distinguish   |  |  |  |  |
|   |  | <ul> <li>between different mutations</li> <li>List the consequences of mutations on the cells, the phenotype, the growth and the development of the living being</li> </ul> |          |   |                       |                               |  |  |  |  |
|   |  |   |          |   |                       |                               |  |  |  |  |
|   |  | <ul> <li>Explain the meaning of different laboratory methods</li> </ul>   |          |   |                       |                               |  |  |  |  |
| 11.   | Content summary:   |   |          |   |                       |                               |  |  |  |  |
|   | Theoretical lessons:   |   |          |   |                       |                               |  |  |  |  |
|   | Basics of human genetics – organization of the prokaryotic and eukaryotic DNA, cell nucleus DNA  |   |          |   |                       |                               |  |  |  |  |
|   | and non-nucleus DNA, basic processes of replication, transcription and translation, regulation of  |   |          |   |                       |                               |  |  |  |  |
|   | genetic expression and signalization, foundations of cytogenetics, chromosomes organization, types   |   |          |   |                       |                               |  |  |  |  |
|   | of chromosomal aberrations, cell and molecular base of inheritance, Mendel's genetics, non-  |   |          |   |                       |                               |  |  |  |  |
| Mendelian, complex and multifactorial inheritance, genetic factors of common disorders. |  |   |          |   |                       |                               |  |  |  |  |
|   |  |   |          |   |                       | ns – types, ways of occurring |  |  |  |  |
|   | and correction syste   |   |          |   |                       |                               |  |  |  |  |
|   |  |   |          |   |                       | nunogenetics. Methods of      |  |  |  |  |
|   | 00   | <b>v</b>  | - and po | stnatal, t  | basic principles of g | enetic counseling, ethical    |  |  |  |  |
|   | aspects of genetic to  | esting.   |          |   |                       |                               |  |  |  |  |
|   | Practical lessons:   |   |          |   |                       |                               |  |  |  |  |
|   |  | f genetic analys  | is – DN  | A extrac  | tion                  |                               |  |  |  |  |
|   |  |   |          |   |                       | alysis of polymorphism        |  |  |  |  |
|   | <ul> <li>Methods of discovering familiar and unfamiliar mutations, analysis of polymorphism</li> <li>Writing and interpreting results in case of morbidity assessment</li> </ul> |   |          |   |                       |                               |  |  |  |  |
|   | -  | ~ -   |          |   | •                     |                               |  |  |  |  |
|   | <ul> <li>Basics of cytogenetics – making a karyotypem staining methods, FISH technique,<br/>recognizing chromosomal aberrations</li> </ul>                                       |   |          |   |                       |                               |  |  |  |  |
|   | <ul> <li>Methods of prenatal and postnatal detection of malformations, genetic counseling</li> </ul>   |   |          |   |                       |                               |  |  |  |  |
| 12.   | Teaching methods:  |   |          |   |                       |                               |  |  |  |  |
| 13.   | Total classes:   | 60  |          |   |                       |                               |  |  |  |  |
| 14.   | Organization   |   | Ī        | 30 theoretical lessons, practical lessons, seminars |                       |                               |  |  |  |  |
|   | -  |   |          | 30 lessons learning at home                         |                       |                               |  |  |  |  |
| 15.   | Types of teaching a  | ctivities   |          | 15.1  | Lessons:              | 15                            |  |  |  |  |
|   |  |   |          |   | theoretical           |                               |  |  |  |  |
|   |  |   | Ļ        |   | classes               |                               |  |  |  |  |
|   |  |   |          | 15.2  | Practical lessons,    | 15                            |  |  |  |  |

|     |                              |  |   | seminars  |                  |                                  |       |        |  |  |
|-----|------------------------------|--|---|---|------------------|----------------------------------|-------|--------|--|--|
| 16. | Other types of activities    |  | 16.1  | Training  |                  |                                  |       |        |  |  |
|     |                              |  | 16.2  |   | ntina            |                                  |       |        |  |  |
|     |                              |  | 16.2  | Self-suppo<br>practice                            | rung             |                                  |       |        |  |  |
|     |                              |  | 16.3  | 1   | 4 1              | 30                               |       |        |  |  |
|     | Vacual des soos              |  | Learning a  | t nome  | 30               |                                  |       |        |  |  |
| 17. | Knowledge assessment         |  | Points  |   |                  |                                  |       |        |  |  |
|     | 17.1                         | Tests  | <b>XX</b> 7   | 6 4 4   | • ,              | 04.40.4                          |       |        |  |  |
|     | 17.2                         | Final exam   | Oral ex   | n form test<br>kam                                | points<br>points | 24-40 (minmax.<br>18-30 (minmax. | ·     |        |  |  |
|     | 17.3                         | Active participation   |   |   |                  | Μ                                | in. – | - Max. |  |  |
|     |                              |  | Theoretical lessons 6 - 10  |   |                  |                                  |       | 10     |  |  |
|     |                              |  | Practical lessons 12  |   |                  |                                  |       | 20     |  |  |
| 18. | Grading                      | Up to 59   | 5 (five) F  |   |                  |                                  |       |        |  |  |
|     | criterion<br>(points/grades) | 60-68  | 6 (six) E   |   |                  |                                  |       |        |  |  |
|     |                              | 69-76  | 7 (seven) D   |   |                  |                                  |       |        |  |  |
|     |                              | 77-84  | 8 (eight) C   |   |                  |                                  |       |        |  |  |
|     |                              | 85-92  | 9 (nine) B  |   |                  |                                  |       |        |  |  |
|     |                              | 93-100   | 10 (ten) A  |   |                  |                                  |       |        |  |  |
| 19. |                              |  |   | ,   |                  | nointe from visitir              | a th  | 2      |  |  |
| 19. | Requirements                 | To obtain a signature the student must gain minimum points from visiting the         |   |   |                  |                                  |       |        |  |  |
|     | for obtaining a              | theoretical and practical lessons.   |   |   |                  |                                  |       |        |  |  |
|     | signature and                |  |   |   |                  |                                  |       |        |  |  |
|     | attending the                | To attend the final (oral) exam, the student must pass the mid-term exams or gain    |   |   |                  |                                  |       |        |  |  |
|     | final                        | at least 30% of the total number of points of the mid-term exams. In the exam        |   |   |                  |                                  |       |        |  |  |
|     | examination                  | session, the student first attends the mid-term exams and then the final exam.       |   |   |                  |                                  |       |        |  |  |
|     |                              |  |   |   |                  |                                  |       |        |  |  |
|     |                              | The final grade for the subject is formed according to the table for grading, and is |   |   |                  |                                  |       |        |  |  |
|     |                              | based on the sum of the points from all the activities, mid-term exams and final     |   |   |                  |                                  |       |        |  |  |
|     |                              | exam.  | _   |   |                  |                                  |       |        |  |  |
| 20. | Language                     | Macedonian   |   |   |                  |                                  |       |        |  |  |
| 21. | Method of                    | Students' anonymous e  | valuation   | n of the subie                                    | ects, the        | professors and colla             | bora  | ators  |  |  |
|     | evaluating the               | who hold the lessons.  |   | j-  | ····, ····       | r                                |       |        |  |  |
|     | quality of the               | who hold the lessons.  |   |   |                  |                                  |       |        |  |  |
|     | lessons                      |  |   |   |                  |                                  |       |        |  |  |
| 22. | Literature:                  |  |   |   |                  |                                  |       |        |  |  |
| 22. |                              |  |   |   |                  |                                  |       |        |  |  |
|     | 22.1                         | Mandatory literature   | Decf 1  | dical Constine U.                                 |                  | : t++ C ~                        |       |        |  |  |
|     |                              | 1.   |   |   |                  | dical Genetics, Uni              |       |        |  |  |
|     |                              | -  |   |   |                  | cal Faculty Skopje,              |       | 3      |  |  |
|     |                              | 2.   | Prof. d-r A. Petlichkovski, Genetics – Authorized                   |   |                  |                                  |       |        |  |  |
|     |                              |  |   | lectures, 2019                                    |                  |                                  |       |        |  |  |
|     |                              | 3.   |   | Prof. d-r M.Spiroski, Human Genetics Practicum 1, |                  |                                  |       |        |  |  |
|     |                              |  | University Ss. Cyril and Methodeus, Medical Faculty                 |   |                  |                                  |       |        |  |  |
|     |                              |  | Skopje. 2009  |   |                  |                                  |       |        |  |  |
|     |                              | 4.   | Prof. d-r Kochova et al., Human Genetics Practicum 2,               |   |                  |                                  |       |        |  |  |
|     |                              |  | University Ss. Cyril and Methodeus, Medical Faculty<br>Skopje, 2009 |   |                  |                                  |       |        |  |  |
|     |                              |  |   |   |                  |                                  |       |        |  |  |
|     | 22.2                         | Additional literature  |   |   |                  |                                  |       |        |  |  |
|     | 44.4                         |  | netics, 3 <sup>rd</sup> ed., Pearson, 2009                          |   |                  |                                  |       |        |  |  |
|     |                              |  | tion and  | ad Dages ar                                       | 2000             |                                  |       |        |  |  |