

1. COURSE DESCRIPTION – GENERAL INFORMATION			
1.1. Course teacher	Professor Karmela Barišić, PhD	1.6. Year of study	4 th
1.2. Name of the course	Molecular Diagnostics	1.7. Credit value (ECTS)	5
1.3. Associate teachers	Marija Grdić Rajković, PhD Andrea Hulina, mag. med. biochem.	1.8. Type of instruction (number of hours L+E+S+e-learning)	30+15+15
1.4. Study programme (undergraduate, graduate, integrated)	Integrated study of Medical biochemistry	1.9. Expected enrolment in the course	25
1.5. Status of the course	Compulsory	1.10. Level of use of e-learning (1, 2, 3 level), percentage of instruction in the course on line (20% maximum)	2 nd
2. COURSE DESCRIPTION			
2.1. Course objectives	Course objectives are to familiarise students with principles and development of molecular diagnostic methods, their use in research, diagnosis and monitoring of diseases.		
2.2. Enrolment requirements and required entry competences for the course	Enrolment requirements: completed Haematology II and Molecular Biology with Genetic Engineering and passed exam in General Clinical Biochemistry.		
2.3. Learning outcomes at the level of the study programme to which the course contributes	<ul style="list-style-type: none"> Defining, analysing and choosing procedures connected to research, manufacturing and quality assurance and implementation of molecular diagnostic procedures for detection and follow-up of medical conditions and efficiency of the therapy. Development and implementation of solutions for practical problems of molecular diagnostics by means of observational, analytical and critical skills. Critical evaluation and implementation of scientific findings and available data in order to improve the field, solving molecular diagnostic problems, implementation of new technologies and improvement of the existing ones. 		
2.4. Expected learning outcomes at the level of the course (4-10 learning outcomes)	<p>On completion of this course, the student will be able to:</p> <ol style="list-style-type: none"> Critically read, interpret and communicate original research literature in molecular diagnostics; Describe techniques used in molecular diagnostics; Explain and practically apply principles and methods used in diagnostics of hereditary diseases; Analyse theoretical and experimental limitations for a defined molecular diagnostic problem; 		

	5. Interpret results of a specific molecular diagnostic test; 6. Describe the ethical aspects in relation to genetic counselling; 7. Define quality criteria for the clinical use of molecular diagnostics; 8. Define the optimal method for a specific molecular diagnostic problem; 9. Use available databases; 10. Participate in the interdisciplinary collaboration with the health sector / health industry on the development of molecular diagnostics.					
2.5. Course content broken down in detail by weekly class schedule (syllabus)	<p>LECTURES:</p> <ul style="list-style-type: none">• Molecular genetics for diagnostics of hereditary diseases (PCR for specific mutation tests, tests for general mutations, DNA sequencing).• Methods for characterization of gene expression (micro arrays and quantitative PCR).• Laboratory management, quality control, validation, variation sources, distinctive property and test sensitivity for molecular diagnostics.• Forensic analyses.• Molecular diagnostics in haematology.• Molecular diagnostics in transfusion medicine.• Molecular methods in epigenome analysis.• Ethical aspects of genetic testing and consultations. <p>SEMINARS:</p> <ul style="list-style-type: none">• Molecular diagnostics in monogenic diseases (cystic fibrosis, Huntington’s disease, Duchenne muscular dystrophy, fragile X chromosome syndrome).• Pharmacogenetics and pharmacogenomics.• Use of available databases on human genome, transcriptome, polymorphisms, genetic variations and diseases. <p>EXERCISES:</p> <ul style="list-style-type: none">• Isolation of nucleic acids, gel electrophoresis, PCR-RFLP analysis, real-time PCR, use of databases, statistical analyses					
2.6. Type of instruction	<u>lectures</u> <u>seminars and workshops</u> <u>exercises</u> online in entirety mixed e-learning field work		<u>independent study</u> multimedia and the internet <u>laboratory</u> work with the mentor (other)		2.7. Comments:	
2.8. Student responsibilities	Regular attendance of all parts of the course, active participation in solving cases from clinical practice, writing the seminar paper (researching literature, essay writing and oral presentation)					
2.9. Screening of student’s work	Class attendance	1.0	Research		Practical training	0.5

(specify the proportion of ECTS credits for each activity so that the total number of CTS credits is equal to the credit value of the course)	Experimental work		Report		Project	
	Essay	1.0	Seminar essay	1,0	(Other--describe)	
	Tests		Oral exam	1.0	(Other—describe)	
	Written exam		Project	0.5	(Other—describe)	
2.10. Grading and evaluation of student work over the course of instruction and at a final exam	Each student's advancement is continually monitored over the course. The final grade comprises overall endeavour of a student during lectures, seminars, exercises and oral exam. Students are required to write a seminar paper which is presented to other students. The seminar paper requires a project approach to a specific topic, an independent research of scientific and specialist literature, writing of the essay, oral presentation along with the discussion on the topic.					
2.11. Required literature (available at the library and via other media)	Title					
	DE Bruns, ER Ashwood, CA Burtis, Fundamentals of Molecular diagnostics, Saunders Elsevier, 2007.					
	Štrausova medicinska biokemija, ur. D. Čvorišćec, I. Čepelak, Medicinska naklada, Zagreb 2009.					
2.12. Optional literature	Materials from lectures and seminars Felgenhauer K Laboratory Diagnosis of Neurological Diseases. In: Thomas L, ed., Clinical Laboratory Diagnostics – Use And Assessment of Clinical Laboratory Results: Frankfurt: TH Books, 1998: 1308-1326					
2.13. Methods of monitoring quality that ensure acquisition of exit competences	Continuous monitoring of students' performance during lectures, seminars, project and essay preparation, solving cases from clinic practice, laboratory performance, evaluation of the presentation (essay) and oral exam. Survey after the end of the course.					