

Molecular Human Genetics						
Identification number	Workload	Credit points	Term of studying	Frequency of occurrence	Duration	
MN-B-SM (G 4)	360 h	12CP	1 st or 2 nd term of studying	Summer term, 2 nd half	7 weeks	
1	Type of lessons		Contact times	Self-study times	Intended group size*	
	a) Lectures		10 h	20 h	max. 9	
	b) Practical/Lab		155 h	136 h	max. 1	
	c) Seminar		15 h	24 h	max. 1	
2	Aims of the module and acquired skills Students who successfully completed this module ... <ul style="list-style-type: none"> • have gained in-depth knowledge in modern human genetics methods. • have acquired experimental skills in state-of-the art molecular genetics and molecular biology (see contents of the module) and can independently design and perform small scientific projects related to the topics of the module. • have learned how to present research results in oral and written form and to critically discuss scientific publications related to the topic of the module on a professional level. • are able to transfer skills acquired in this module to other fields of biology. 					
3	Contents of the module <ul style="list-style-type: none"> • Identification and characterization of the molecular basis of human inherited diseases (neuromuscular and neurodegenerative disorders, kidney diseases, , skeletal disorders, and hereditary tumor predisposition syndromes) and of rare developmental syndromes. Subtopics: disease gene location (linkage studies), identification of disease genes (targeted (Panel) and whole exome sequencing using next generation sequencing), identification of underlying mutations, functional analysis of disease genes in vitro and in vivo, functional analysis of the disease relevant protein complexes • Identification of disease modifying/protective factors • Therapeutic approaches (pharmacotherapy, epigenetic approaches, gene therapy) • Molecular genetic technologies (PCR, sequencing, real-time PCR, genotyping of polymorphic markers, RT-PCR, pyrosequencing, Southern-blotting, etc.) • Analysis of sequencing data and mutations, construction of haplotypes, construction of primers, assembling and alignment of sequences, etc. • Molecular cloning (cloning of PCR fragments into plasmids, isolation of plasmid DNA, transfections); use of CRISPR/Cas-system • Cell culture technology (working with human and murine cell lines) • Immunohistochemistry, fluorescence microscopy • Protein analysis and protein-interaction methods (Western blotting, co-immunoprecipitation of proteins, pull-down, chromatin-immunoprecipitations (ChIP) etc.) • Analysis of knock-out and transgenic mice and zebrafish <p><i>Explanatory note:</i> The list above comprises topics and techniques that are commonly used at the Institute of Human Genetics, CECAD, CCG, Epigenomics and Experimental Immunology of the Eye. Thus, every student participating in this module will be confronted with a large subset of it. The exact content, however, will depend on the tutor and the research project the student will work on.</p>					
4	Teaching/Learning methods <ul style="list-style-type: none"> • Lectures; Practical/Lab (Project work); Seminar; Computer exercises; Guidance to independent research; Training on presentation techniques in oral and written form 					

5	<p>Requirements for participation</p> <p>Enrollment in the Master´s degree course "Biological Sciences" or in the Master´s degree course "Biochemistry"</p>
6	<p>Type of module examinations</p> <p>The final examination consists of three parts: 30 min oral examination about topics of the lectures and the practical/lab part (50 % of the total module mark), oral presentation (25 % of the total module mark) and seminar paper (25 % of the total module mark)</p>
7	<p>Requisites for the allocation of credits</p> <p>Regular and active participation; Each examination part at least "sufficient" (see appendix of the examination regulations for details)</p>
8	<p>Compatibility with other Curricula</p> <p>Biochemical subject module in the Master´s degree course "Biochemistry"</p>
9	<p>Significance of the module mark for the overall grade</p> <p>In the Master´s degree course "Biological Sciences": 15 % of the overall grade (see also appendix of the examination regulations)</p>
10	<p>Module coordinator</p> <p>Prof. Dr. BrunhildeWirth, phone 478-86464, e-mail: brunhilde.wirth@uk-koeln.de</p>
11	<p>Additional information</p> <p>Subject module of the Master´s degree course "Biological Sciences", Focus of research: (G) Genetics and Cell Biology</p> <p>Participating faculty: Dr. B. Beck, Prof. Dr. M. Bergami, Dr. R. Hansel-Hertsch, Dr. N. Kononenko, Prof. Dr. T. Langmann, Dr. N. Mendoza-Ferreira, Prof. Dr. P. Nürnberg, Prof. Dr. M. Schweiger, Prof. Dr. B. Wirth, Dr. H. Zempel</p> <p>Location: The lab part will be held depending on the PI at 1) Center for Molecular Medicine Cologne, 2) CECAD, 3) Department of Ophthalmology or 4) CCG, 50931 Cologne. Seminars will be held at the Institute of Human Genetics, library (Frauenklinik Building 47, Kerpener Str. 34, ninth floor)</p> <p>Literature:</p> <ul style="list-style-type: none"> • Strachan, T., Read, A.P. (2019) Human Molecular Genetics. 5th edition, Garland Science • Nussbaum, R.L., Willard, H.F., McInnes, R.R. (2015) Thompson and Thompson - Genetics in Medicine. 8th edition, Saunders • For those students, who speak German: Hirsch-Kauffmann, M., Schweiger, M., Schweiger, M.R. (2009) Biologie und Molekulare Medizin. 7. Auflage, Thieme <p>General time schedule: Week 1-6 (Mon.-Fri.): Lectures, practical/lab and preparation for the seminar talk (topic and date will be arranged individually) as well as writing seminar paper; Week 7 (Mon.-Fri): Preparation for the oral examination</p> <p>Note: The module contains hand-on laboratory work conducted individually and is taught in research laboratories. The module does not contain computer-based practicals/ research as a main component.</p> <p>Introduction to the module: May 20, 2020 at 4:15 p.m., Institute of Human Genetics, library (Frauenklinik Building 47, Kerpener Str. 34, ninth floor)</p> <p>Oral examination: July 17, 2020, second/supplementary examination August 28, 2020; the latter date may vary if students and module coordinator agree. More details will be given at the beginning of the module.</p>

*5 students from the Master´s degree course "Biological Sciences" and 4 students from the Master´s degree course "Biochemistry".