

<b>Module Name</b> Molecular Human Genetics						
<b>Type of Module</b> ○ Advanced Module				<b>Module Code</b> Human Genetics		
<b>Identification Number</b> MN-B-SM (Z 3)	<b>Workload</b> 360 h	<b>Credit Points</b> 12 CP	<b>Term</b> 2 <sup>nd</sup> term of studying	<b>Offered Every</b> Summer term	<b>Start</b> summer term only	<b>Duration</b> 7 weeks
<b>1</b>	<b>Course Types</b> a) Lectures b) Practical/Lab c) Seminar		<b>Contact Time</b> 10 h 155 h 15 h	<b>Private Study</b> 20 h 136 h 24 h	<b>Planned Group Size*</b> max. 8 max. 1 max. 1	
<b>2</b>	<b>Module Objectives and Skills to be Acquired</b> Students who successfully completed this module <ul style="list-style-type: none"> <li>• have gained in-depth knowledge in modern human genetics methods.</li> <li>• 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.</li> <li>• 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.</li> <li>• are able to transfer skills acquired in this module to other fields of biology.</li> </ul>					

<p><b>3</b></p>	<p><b>Module Content</b></p> <ul style="list-style-type: none"> <li>• 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</li> <li>• Identification of disease modifying/protective factors</li> <li>• Therapeutic approaches (pharmacotherapy, epigenetic approaches, gene therapy)</li> <li>• Molecular genetic technologies (PCR, sequencing, real-time PCR, genotyping of polymorphic markers, RT-PCR, pyrosequencing, Southern-blotting, etc.)</li> <li>• Analysis of sequencing data and mutations, construction of haplotypes, construction of primers, assembling and alignment of sequences, etc.</li> <li>• Molecular cloning (cloning of PCR fragments into plasmids, isolation of plasmid DNA, transfections); use of CRISPR/Cas-system</li> <li>• Cell culture technology (working with human and murine cell lines)</li> <li>• Working with inducible pluripotent stem cells (iPSC) and neuronal differentiation</li> <li>• Immunohistochemistry, fluorescence microscopy</li> <li>• Protein analysis and protein-interaction methods (Western blotting, co-immunoprecipitation of proteins, pull-down, chromatin-immunoprecipitations (ChIP) etc.)</li> <li>• Analysis of knock-out and transgenic mice</li> <li>• <i>Explanatory note:</i> The list above comprises topics and techniques that are commonly used at the Institute of Human Genetics, CECAD, CMMC, 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.</li> </ul>
<p><b>4</b></p>	<p><b>Teaching Methods</b></p> <p>Lectures; Practical/Lab (Project work); Seminar; Computer exercises; Guidance to independent research; Training on presentation techniques in oral and written form</p>
<p><b>5</b></p>	<p><b>Prerequisites (for the Module)</b></p> <p>Enrollment in the Master´s degree course “Biological Sciences”, in the Master’s degree course “Biochemistry”</p>
<p><b>6</b></p>	<p><b>Type of Examination</b></p> <p>The final examination consists of two parts: oral examination on topics of lectures, seminars and the practical/lab part (20-30 min; 50 % of the total module mark) written report (50 % of the total module mark)</p>
<p><b>7</b></p>	<p><b>Credits Awarded</b></p> <p>Regular and active participation Each examination part at least “sufficient” (see appendix of the examination regulations for details)</p>
<p><b>8</b></p>	<p><b>Compatibility with other Curricula*</b></p> <p>Biochemical subject module in the Master´s degree course “Biochemistry”</p>

9	<p><b>Proportion of Final Grade</b></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><b>Module Coordinator</b></p> <p>Prof. Dr. Brunhilde Wirth, phone 478-86464, e-mail: brunhilde.wirth@uk-koeln.de</p>
11	<p><b>Further Information</b></p> <p><b>Subject module</b> of the Master's degree course "Biological Sciences",</p> <p><b>Participating faculty:</b> PD Dr. B. Beck, Prof. Dr. M. Bergami, Dr. R. Hänsel-Hertsch, Prof. Dr. N. Kononenko, Prof. Dr. T. Langmann, Prof. Dr. M. Schweiger, Prof. Dr. B. Wirth, Dr. H. Zempel</p> <p><b>Location:</b> 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><b>Literature:</b></p> <ul style="list-style-type: none"> <li>• Strachan, T., Read, A.P. (2019) Human Molecular Genetics. 5<sup>th</sup> edition, Garland Science</li> <li>• Nussbaum, R.L., Willard, H.F., McInnes, R.R. (2015) Thompson and Thompson - Genetics in Medicine. 8<sup>th</sup> edition, Saunders</li> <li>• For those students, who speak German: Hirsch-Kauffmann, M., Schweiger, M., Schweiger, M.R. (2009) Biologie und Molekulare Medizin. 7. Auflage, Thieme</li> </ul> <p><b>General time schedule:</b> 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><b>Note:</b> 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><b>Introduction to the module:</b> May 18, 2022 at 4:15 p.m., Institute of Human Genetics, library (Frauenklinik Building 47, Kerpener Str. 34, ninth floor)</p> <p><b>Oral examination:</b> July 15, 2022, second/supplementary examination August 26, 2022; the latter date may vary if students and module coordinator agree. More details will be given at the beginning of the module.</p>

\* 4 students from the Master's degree course "Biological Sciences" and 4 students from the Master's degree course "Biochemistry".