Final Exam Human Genetics – Modul BIO-M-TM-HG

In preparation for your final exam ("Modulprüfung") in Human Genetics….

1) Make sure that you have completed all seminars, lab courses etc. required for admission to the final exam.
2) Choose one topic for a PPT presentation from the list below.
3) Contact the office of the Institute of human Genetics, Mrs. Regina Gellner (regina.gellner@klinik.uni-regensburg.de), to schedule an appointment for the oral exam, let her know the topic your choice.
4) Confirm time and date with Mrs. Gellner and Mrs. Lang from the "Zentrales Prüfungssekretariat - Prüfungsamt Biologie / Biochemie".
5) The exam will start with your **10 minute** (15 min max.!!!) PPT presentation. The slides need to be in English, the actual presentation can be inEnglish or German. Your talk will be followed by **30 minutes of questions** around your topic (and related subjects).
6) On the day of your exam: Come to the Institute of Human Genetics, University Clinic, Building D3, Level 5 and see Mrs. Gellner about 15 minutes before your exam.

If required, we can provide a laptop (Windows PC, Mac users are welcome to bring their own!) and you will give your talk in Prof. Weber's office (building D3, room 5.13). The minutes will be recorded.

Please note: Even though M.Sc. Biology students do not have to take the written exam on the lecture "Advanced Human Genetics", all topics taught in this lecture are relevant for the final exam!

Topics:

1) Fragile X-Syndrome mechanisms of disease and therapeutic approaches
2) : Muscular Dystrophies Duchenne and Becker
3) Trinucleotide repeat diseases - mechanisms of disease and therapeutic approaches
4) "Short/long RNAs" and their involvement in gene regulation
5) Stargardt Disease - approaches to therapy
6) The innate immune system and its role in age related macular degeneration
7) Gene identification in complex diseases – the example of age-related macular degeneration
8) Molecular basis of tumor development – latest advances and consequences for treatment
9) Familiar tumor predisposition – the example of hereditary breast and ovarian cancer
10) CRISPR/Cas9 mediated gene editing and its potential application from bench to bedside
11) Adeno-associated virus based gene therapy in the clinic – possibilities, problems and perspectives
12) Copy Number Variation and its relevance in healthy and disease
13) Personalized Medicine – Anti-cancer therapy using Olaparib
14) X-chromosome inactivation in females: molecular mechanisms and clinical relevance

15) Cystic Fibrosis – from the molecular understanding of pathology to therapeutic approaches
16) Cryptic splice sites and their involvement in hereditary retinal dystrophies
17) Gene identification by positional cloning
18) Genome medicine – status quo in Europe and US
19) New advances in long-range sequencing technology
20) Chromosome or gene therapy in Trisomie 21 - practical and ethical challenges