Curriculum for 4-weeks education in molecular genetics of haemophilia

Malmö Center for thrombosis and haemostasis has an “in-house” combined routine and research laboratory for molecular genetics of haemophilia A and B as well as von Willenbrand disease. The laboratory can perform all available molecular diagnostic procedures for the F8, F9 and VWF genes such as, sequencing, analysis of different inversions in the F8 gene, linkage analysis, MLPA and as part of research projects also haplotyping of the genes, NGS (next generation sequencing) and studies of mRNA by RT-PCR and sequencing of cDNA products. Thus the laboratory performs genotyping of patients, carrier- and prenatal diagnosis.

Aim of curriculum

The student will get an overview of the methods used for molecular diagnosis of haemophilia and VWD and how genetic information is interpreted and applied in the clinical care of patients.

Theoretical curriculum

Before the course the student’s background knowledge will be discussed in order to individualize the education. Depending on the knowledge, relevant literature will be provided as well as discussions of current knowledge from large international multi-center studies, suitable public websites with relevant genetic information and “mini-workshops” together with the supervisor. Topics to be covered are:

- The structure of the F8 and F9 genes
- Overview of the techniques used for sequencing and linkage analysis using polymorphisms
- Laboratory pitfalls
- The requirements for good quality/accreditation of the methods
- Clinical, ethical and psychological aspects of carrier- and prenatal diagnosis
- VWF protein structure-function relationship
- VWF gene organization and genetic alterations of the VWF gene causing VWD.
- Cases, illustrating common pitfalls in VWF phenotyping.

Practical curriculum

The student will have “hands-on” in the laboratory under the guidance of a laboratory technician and follow the laboratory process from DNA-extraction to computerized sequence data. The student will participate in the evaluation and clinical interpretation of results of molecular genetic analysis and tasks will involve searching the web for genetic information and evaluate the certainty that a given mutation of the genes will be causative of the disorder. Educational examples of carrier- and prenatal diagnosis and well as genotyping of haemophilia patients from practice will be presented to the student for evaluation and interpretation and then for discussion with the supervisor. The educational samples will illustrate the possibilities and limitations with current techniques and knowledge of the human genome.
Depending on the student’s background and interest in research, possibilities will be given to be introduced to on-going research projects in the field.

*Faculty*

The course will be led by MDs with expertise in molecular genetics and clinical care of haemophilia and an experienced laboratory technician.