DEPARTMENT OF MOLECULAR MEDICINE AND SURGERY

K1F2981, Rare Disease Genomics, 1.5 credits (hec)

Genomik för sällsynta sjukdomar, 1,5 högskolepoäng

Third-cycle level / Forskarnivå

Approval

This syllabus was approved by The Committee for Doctoral Education on 2023-11-13, and was last revised on 2025-09-13. The revised course syllabus is valid from spring semester 2026.

Responsible department

Department of Molecular Medicine and Surgery, Faculty of Medicine

Prerequisite courses, or equivalent

No prerequisite courses, or equivalent, demanded for this course.

Purpose & Intended learning outcomes

Purpose

This is a course aimed at students actively involved or planning genetic analysis of rare diseases. The course is also appropriate for those working with complex diseases and cancer whose projects involve high throughput sequencing. The purpose of this course is to provide the participants with knowledge and practical experience about current research strategies and tools for analysis of sequencing data in the field of rare disease genomics. The participants will also be made aware of ethical issues in relation to rare disease genomics.

Intended learning outcomes

After the course, the participants should be able:

1. To select adequate sequencing technologies and data analysis strategies to answer research questions in the field of rare disease genetics or in their field of research;

2. To evaluate candidate variants and genes using publicly available databases and tools;

3. To discuss suitable approaches for functional validation of candidate variants and genes;

4. To identify and discuss on ethical issues arising from large-scale sequencing.

Course content

The focus of the course is the use of current sequencing methods and bioinformatics tools to understand the basis of rare genetic diseases. Within the overall theme of clinical and experimental approaches to diagnostics of rare genetic diseases, particular attention will be paid to annotation and interpretation of different types of genetic variants. The course will cover the use of different in-silico pathogenicity scores, systematic deep phenotyping, and population and family data for variant and gene interpretation. The course will cover selected experimental strategies to validate genetic findings. The course will also cover current clinical best practice guidelines concerning ethical issues such as report of incidental findings and acquisition of informed consent.

Forms of teaching and learning

The course consists of lectures, seminars, workshops, and self-studies. Students are required to bring their laptops with working internet connection.

Language of instruction

The course is given in English

Grading scale

Pass (G) /Fail (U)

Compulsory components & forms of assessment

Compulsory components

All teaching and learning activities are compulsory. Absence from compulsory parts is compensated according to the instructions from the course leader.

Forms of assessment

The examination consists of a group discussion and presentation. Each student needs to show that all intended learning outcomes have been achieved to pass the course.

Course literature

Reference literature:

- Wright CF et al. Paediatric genomics: diagnosing rare disease in children. Nat Rev Genet. 2018 May;19(5):253-268.
- Tesi B, Boileau C, Boycott KM, et al. Precision medicine in rare diseases: What is next?. J Intern Med. 2023;294(4):397-412.
- Eisfeldt J, Ek M, Nordenskjöld M, Lindstrand A. Toward clinical long-read genome sequencing for rare diseases. Nat Genet. 2025;57(6):1334-1343.
- Miller DT, Lee K, Abul-Husn NS, et al. ACMG SF v3.2 list for reporting of secondary

findings in clinical exome and genome sequencing: A policy statement of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2023;25(8):100866.

- Gudmundsson S, Singer-Berk M, Watts NA, et al. Variant interpretation using population databases: Lessons from gnomAD. Hum Mutat. 2022;43(8):1012-1030.
- Lappalainen T, MacArthur DG. From variant to function in human disease genetics. Science. 2021;373(6562):1464-1468.