# Course: Medical Genetics — credits: 5

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| **Course code** | BOVH24MDMEDGEN |
| **Name** | Medical Genetics |
| **Study year** | 2025-2026 |
| **ECTS credits** | 5 |
| **Language** | Dutch, with parts in English |
| **Coordinator** | S.A.B. Hermsen |
| **Modes of delivery** | Assignment<br>Lecture<br>Tutorial |
| **Assessments** | Bio-Informatica - Other assessment<br>Medische Genetica - Computer, organised by STAD examinations |

**Learning outcomes**

You explain and draw how mitotic and meiotic division proceeds in human cells and reason about the consequences of nondisjunction of chromosomes during this division as the basis of medical genetics.

You will explain the various forms of aneuploidy (chromosomal abnormalities) and chromosome mutations and reason how they are inherited in humans giving examples of disorders that may result.

You understand epigenetic concepts and genomic imprinting and apply these concepts to explain why they may affect the development of inherited disorders.

You explain and apply the concepts of allele frequency and population genetics in medical genetics issues, relating them to factors that influence them and using calculations to predict how often these disorders occur in a population.

You work out a case, in which you analyze a genealogical pedigree for the purpose of (pre-)diagnostic research of hereditary diseases, analyze and interpret the results of a number of molecular biological techniques used in medical genetics using basic bioinformatics as part of working out a case.

**Content**

Medical genetics is a specialty within medicine that deals with the diagnosis and manageability of inherited disorders. With current developments in human genetics, more and more hereditary diseases can be detected. Within this module, you will learn how inherited disorders arise and how to detect them.

You will begin with a brief review of mitosis and meiosis and learn how they arise and what consequences they have. Then you will look at mutations and chromosomal abnormalities and how they arise. A number of examples will be covered, where you will find out how they can be inherited.

You will also learn about different techniques, such as (next generation) sequencing and karyotyping, used in diagnostics. You will analyze results from these techniques using bioinformatics, covering both the techniques and the analysis in silico. You will use the results to independently solve a case.

**Included in programme(s)**

Major Medical Diagnostics

**School(s)**

Institute for Life Science & Technology

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