



# BB2470 Genetik och genomik

## 10,0 hp

Genetics and Genomics

### Fastställande

Kursplan för BB2470 gäller från och med HT11

### Betygsskala

A, B, C, D, E, FX, F

### Utbildningsnivå

Avancerad nivå

### Huvudområden

Bioteknik

### Särskild behörighet

At least 150 credits from grades 1, 2 and 3 of which at least 100 credits from years 1 and 2, and bachelor's work must be completed. The 150 credits should include a minimum of 20 credits within the fields of Mathematics, Numerical Analysis and Computer Sciences, 5 of these must be within the fields of Numerical Analysis and Computer Sciences, 20 credits of Chemistry, possibly including courses in Chemical Measuring Techniques and 20 credits of Biotechnology or Molecular Biology.

### Undervisningsspråk

Undervisningsspråk anges i kurstillfällesinformationen i kurs- och programkatalogen.

## Lärandemål

### Part one

**Following completion and passing the course you should be able to describe:**

“ the architecture and function of the genomes of the different organisms, and explain how differences and similarities have evolved since the origins of life

“ how genes function and are inherited in different types of organisms, and how this affects the organisms’ function and defence against genetic defects, and their evolution

“ how DNA gets damaged and is repaired in the cells, and how genetic variation, resulting from inheritance or from “fresh” DNA damage, affects our health

“ how genetic variation, among genes, individuals, populations or species, originates and evolves

### Part two

**Following completion and passing the course you should be able to:**

**I. Describe, illustrate and relate different techniques in the fields of genomics and transcriptomics**

**II. Critically evaluate, select and apply the most appropriate technique(s) in different biological and medical studies.**

**III. Discuss and suggest strategies to tackle and solve challenging problems in various research studies.**

**IV. Construct and create biologically relevant studies by employing one or more of the discussed tools.**

## Kursinnehåll

### Part one

Genetics is the basis for most biological, medical and biotechnical analyses and techniques. Consequently, in order to optimally exploit the biotechnical tools, knowledge about the basic genetics is of great importance. This part of the course aims to give a broad knowledge, from an evolutionary perspective, of how genetic variation is formed and inherited, and how it evolves.

A number of basic aspects of genetics will be studied, for example:

The origins of life, and the “Tree of Life”: the origins, development and relationships (phylogeny) of all organisms

The mechanisms of evolution

The genetic difference between organisms: differences and similarities in the architecture and function of the genomes, and how this evolved through the evolution

Inheritance of genes and traits: different modes of inheritance (e.g. Mendelian and asexual) and their effect on the “success” of individuals and species

Inherited diseases: their causes and effects

Mutations: the chemistry of DNA damage, the types of mutations, and cellular mechanisms for their repair

Mapping of genes (identification of which trait is affected by which gene)

Genetic differences between human populations: their historical origin and subsequent spread, and their medical importance

### **Course content: Part two**

This part of the course aims to give detailed insight into the techniques and technological trends in the fields of genomics and transcriptomics, to build up the necessary foundations for further understanding of association studies, pharmacogenomics, forensics, population genetics, diagnostics, medicine and drug development. The course includes a short introduction to conventional assays used in molecular biotechnology, description of different methods for typing of genetic variations, a variety of techniques for multiplex amplification, advanced techniques and platforms for DNA sequencing and whole genome sequencing, and different techniques for transcript profiling.

In addition, the course involves a literature workshop of selected articles, which will be performed in groups. Each group presents one article and will oppose two other groups' articles. This project aims to learn critical reading, interpretation and comparison of the most advanced techniques and platforms in the fields of whole genome sequencing, massive parallel genotyping and transcript profiling. The project requires teamwork and planning, and participation as well as presence on the workshop days is compulsory. The course also includes assignments that should be prepared after each lecture.

## **Kurslitteratur**

### **Part one**

Course book: "Fundamental Genetics" by John Ringo Cambridge University Press 2004

Additional texts: Eleven articles – Reviews, popular science, and book chapters.

### **Part two**

Distributed handouts and articles.

## **Examination**

- PRO1 - Projekt, 1,0 hp, betygsskala: P, F

- TEN2 - Skriftlig tentamen, 4,5 hp, betygsskala: A, B, C, D, E, FX, F
- TEN1 - Skriftlig tentamen, 4,5 hp, betygsskala: A, B, C, D, E, FX, F

Examinator beslutar, baserat på rekommendation från KTH:s handläggare av stöd till studenter med funktionsnedsättning, om eventuell anpassad examination för studenter med dokumenterad, varaktig funktionsnedsättning.

Examinator får medge annan examinationsform vid omexamination av enskilda studenter.

När kurs inte längre ges har student möjlighet att examineras under ytterligare två läsår.

### **Part one**

Written exam (TEN1, 4.5 hp, grade scale A-F).

Account of the additional texts.

### **Part two**

The examination will be based on: Lectures, Handouts and Distributed articles. The written examination will have short essay type of questions (TEN2, 4.5 hp, grade scale A-F).

Project PRO1 (P/F): The project requires teamwork and planning, and participation as well as presence on the workshop days (October 5, 12, 14 and 16) is compulsory.

## **Övriga krav för slutbetyg**

Passing grade on all four parts. Active participation in the literature projects, assignments and written examination.

## **Etiskt förhållningssätt**

- Vid grupparbete har alla i gruppen ansvar för gruppens arbete.
- Vid examination ska varje student ärligt redovisa hjälp som erhållits och källor som använts.
- Vid muntlig examination ska varje student kunna redogöra för hela uppgiften och hela lösningen.