



# BB2470 Genetik och genomik

## 10,0 hp

Genetics and Genomics

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

### Fastställande

Kursplan för BB2470 gäller från och med HT10

### Betygsskala

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

### Utbildningsnivå

Avancerad nivå

### Huvudområden

Bioteknik

### Särskild behörighet

### 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 describe:**

- “ how genome sequencing is performed
- “ conventional methods and platforms for discovery and typing of mutations and single nucleotide polymorphisms (SNPs)
- “ obstacles with multiplex PCR amplification
- “ different techniques and platforms enabling multiplex PCR amplification
- “ different techniques and platforms enabling whole genome SNP genotyping.
- “ conventional methods for DNA sequencing, and also be able to understand limitations with these techniques
- “ most recent developed techniques and platforms for whole genome sequencing and future trends in the field
- “ different microarray-based techniques for parallel analysis of gene expression and also be able to describe experimental design and difficulties with microarray-based approaches
- “ alternative techniques and platforms for analysis of gene expression

In addition to be able to describe these techniques, you should know the application area(s) of each specific approach and should have understanding for advantages and disadvantages of each technique, and also be able to compare and discuss usefulness of the techniques in each field.

## **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

## **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 (October 5, 12, 14 and 16) is compulsory.

## **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
- TEN1 - Skriftlig tentamen, 4,5 hp, betygsskala: A, B, C, D, E, FX, F
- TEN2 - 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 enstaka studenter.

## **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 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.