# Examination

**Course: Human genetics and Applied Bioinformatics**

**Course code: MC2001**

**Course coordinator: Ignacio Rangel**

**Date: 2013-08-13**

**Exam time: 5 h**

**Total points: 67**

**Pass 60 % of total points**
**Pass with Distinction 85 % of total points**

**Answer the questions from each teacher on separate papers and put the papers in a green cover, one cover for each teacher**

**Write code on each sheet of paper**
**Write only on one side of the paper**

***Good Luck!***

**Questions from: Elisabet Tina**

Introduction to Human genetics

1. Define euploid, aneuploid, deltion and amplification. (2 p)
2. Describe three different types of mutations and theirs consequences on the protein sequence and function. (6 p)
3. Explain the event non-disjunction and its consequence for the chromosome set. (2 p)

**Questions from: Robert Kruse**

Microarray

1. Give a detailed description of the principle design of an oligo DNA-microarray and of the hybridisation of target probes during analysis. (2 p)
2. Briefly describe the different microarray applications: (4 p)
    1. Chromatin immunoprecipitation (Chip)
    2. Gene expression profiling (WholeGenome)
    3. Single nucleotide polymorphism (SNP) detection
    4. DNA methylation

## Questions from: Igor Oliynyk

1. Name at least four clinical symptoms associated with Trisomy 13 (Patau syndrome). (2p).

2. What abnormalities of chromosome structure do you know? Give at least four examples.(2p)

3. Fluorescence in situ hybridization (FISH) is often used to identify the presence, absence or rearrangement of DNA segments. Describe the three main stages of the method. (3 p).

4. What does non-disjunction mean and what could be the result of this? (2p)

5. How Cystic Fibrosis is inherited? (1p)

**Questions from: Marike Gabrielsson**

Messenger RNA regulation

1. Describe 2 mechanisms of mRNA regulation (4p)
2. What is a chaperone protein? Describe its function and give an example of one. (2p)

Population genetics

1. Define the following words: *Phenotype*, *Haplotype*, *Heterozygous* and *Oncogene*. (4p)
2. Describe 3 mechanisms involved in population genetics, i.e. mechanisms participating in development or driving force of population genetics. (6p)

**Questions from: Benjamin Ulfenborg**

1. Describe the relationship between a protein's structure and function. (1)
2. Explain what is meant by a misfolded protein and give an example of how misfolding can occur. (2)
3. Describe what amyloid fibrils are, why they occur and how they can cause damage to the body. (3)

**Questions from: Sanja Farkas**

1. Define euchromatin and heterochromatin. (1p)
2. Describe concisely the three major epigenetic mechanisms? (3)
3. Describe the method methylation specific PCR (MSP). (3p)

**Questions from: Mats Karlsson**

1. World medical association has agreed on a declaration on Ethical Principles for Medical Research Involving Human Subjects, name it. 1p.

2. In order to perform a study on human subjects, what kind of body (committee) do you need the permit from? 1p

3. In Sweden, there is a special legislation regulating the clinical and research use of human biological material, what is the name of that legislation? 1p

4. In which of the following situations do you need a permit according to the above mentioned legislation (question 3), answer yes or no for each example. 3p

    - Blood pressure measurement in a study of hypertensive subject

    - A muscle biopsy taken at a study you perform at the department of clinical physiology at Örebro University Hospital

    - A blood sample that you take in a study initiated by Örebro University on a study site in Africa

**Questions from: Allan Sirsjö**

1. Give example of strategy/strategies to investigate the importance of SNPs for the development of complex diseases (cardiovascular diseases). (3p)

2. Give example of factors that could influence genetic tests to predict risk to develop cardiovascular diseases. (3p)