Re-examination

Course: Human genetics and Applied Bioinformatics

Course code: MC2001

Course coordinator: Ignacio Rangel

Date: 2014-06-04

Exam time: 5 h

Total points: 52

Pass 60 % of total points (31p)
Pass with Distinction 85 % of total points (44p)

Answer the questions from each section on separate papers and place the papers in a green cover, one cover for each section

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

Good Luck!

Section 1

1. Define euploid, aneuploid, deletion and amplification. **(2 p)**
2. Describe three different types of mutations and their consequences on the protein sequence and function. **(6 p)**
3. Explain the event non-disjunction and its consequence for the chromosome set. **(2 p)**
4. Give a detailed description of the principle design of an oligo DNA-microarray and of the hybridisation of target probes during analysis. **(2 p)**

Section 2

5. Name at least four clinical symptoms associated with Trisomy 13 (Patau syndrome). **(2p)**
6. 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)**
7. How Cystic Fibrosis is inherited? **(1p)**

Section 3

8. Describe 2 mechanisms of mRNA regulation **(4p)**
9. What is a chaperone protein? Describe its function and give an example of one. **(2p)**
10. Define the following words: *Phenotype*, *Haplotype, Heterozygous* and *Oncogene*. **(4p)**
11. Describe 3 mechanisms involved in population genetics, i.e. mechanisms participating in development or driving force of population genetics. **(6p)**
12. Describe the relationship between a protein's structure and function. **(1p)**
13. Explain what is meant by a misfolded protein and give an example of how misfolding can occur. **(2p)**
14. Describe concisely the three major epigenetic mechanisms? **(3p)**

Section 4

15. World medical association has agreed on a declaration on Ethical Principles for Medical Research Involving Human Subjects, name it. **(1p)**
16. In order to perform a study on human subjects, what kind of body (committee) do you need the permit from? **(1p)**
17. 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)**
18. 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)**
    a. Blood pressure measurement in a study of hypertensive subject

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

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

Section 5

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

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