NTNU

Faculty of Medicine
Department of Laboratory Medicine, Children's and Women's Health

Exam MOL3001
Medical genetics

Monday 30 May 2011, 9.00 am - 1.00 pm

ECTS credits: 7.5
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Examination support: Calculator and English dictionary

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Examination results are announced on http://studweb.ntnu.no/

All questions are equally rated (25% for each of question 1-4)

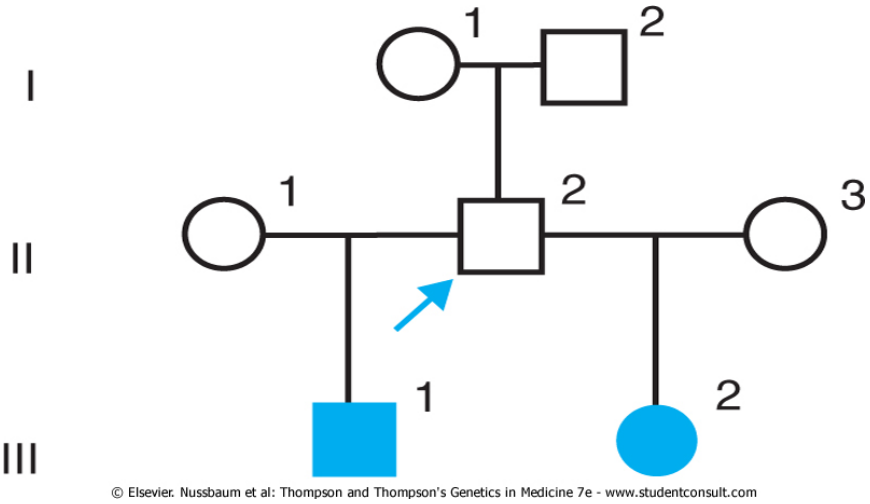
Examination question 1.

Monogenic diseases are often named Mendelian disorders.

a) Describe what is meant by Mendelian inheritance, and give examples of different inheritance patterns.

b) Factors that can affect inheritance patters are penetrance and expressivity. Explain these terms and describe how these factors may influence the inheritance patterns.

c) How can you explain the inheritance of the disease in the pedigree below?



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Examination question 2.

a) Explain the concept of genomic imprinting. Use examples of diseases associated with genomic imprinting to illustrate your answer.

b) Eva is 25 years old and has had three children with Down syndrome. Give two possible explanations for this.

c) Explain why chromosomal aberrations rarely are propagated to the next generation?

Examination question 3.

a) Twin-studies are often employed in order to separate the contribution of genetic from environmental influences in multifactorial diseases. For diseases with a significant genetic component, the concordance rate in monozygotic (MZ) twins is generally higher than in dizygotic (DZ) twins. Explain why.

b) For a complex trait, the concordance rates in MZ and DZ twins were found to be 30% and 25%, respectively. What do these concordance rates tell about the contribution from genetic and environmental factors to the development of the trait?

c) In a comprehensive linkage analysis of a recessive autosomal disorder, an informative genetic marker was found to co-segregate with the disease in about 50% of the cases. What can you say about the location of the disease gene relative to the genetic marker?

Examination question 4.

a) The best known genetic risk factor for sporadic Alzheimer's disease (AD) is the inheritance of the Apolipoprotein E ε4 allele (*APOE* ε4), and between 40 and 80 % of patients with AD possess at least one ε4 allele. However, 50-70% of heterozygotes for the ε4 allele never develop AD. What do these numbers indicate about the etiology of AD?

b) Many monogenic diseases are caused by a gene defect leading to an enzyme deficiency. Describe possible consequences of an inherited enzyme defect, preferable with an example.

c) Discuss ethical implications of termination of pregnancies due to fetal aberrations.

Made by Marit Anthonsen, Frank Skorpen, Inga Bjørnevoll og Wenche Sjursen