

Statistical Methods in Genetics, fall 2008

Examn - January 22th, 2009

Your Name :

Student id :

Question [1]: Explain shortly the following terms. **(6 points)**

- (a) Penetrance probability.
- (b) IBD and IBS.
- (c) Genetic map-function.
- (d) Genetic heterogeneity.
- (e) Linkage disequilibrium (LD).
- (f) Hardy-Weinberg equilibrium (HWE).

Question [2]: Formulate the mixed model (= genetic term) which is also sometimes called mixed inheritance model. Explain the terms (effects) in the model and state the typical assumptions. **(6 points)**

Question [3]: Explain with your own words what are the basic ideas in linkage analysis and in association analysis. Try to explain what differences there are. Statistical details can be kept to a minimum. **(6 points)**

Question [4]: Let us have a pedigree data and the markers (M_1, M_2, M_3, M_4, M_5) with recombination fractions ($\theta_1, \theta_2, \theta_3, \theta_4$) between markers. When we place the trait locus l on the position between the markers M_3 and M_4 , we obtain recombination fractions θ_L (between M_3 and the trait locus l) and θ_R (between trait locus l and M_4). Please explain then shortly how can we calculate the multipoint likelihood (for these markers and the trait locus l jointly) using the Lander-Green algorithm. **(6 points)**

Question [5]: Outline with your own words the basic ideas in the Elston-Stewart peeling algorithm. Explain, to what purpose it is intended for, what are its good properties and what limitations it has. **(6 points)**

Statistical Methods in Genetics, fall 2008

Examn - April 2th, 2009

Your Name :

Student id :

Question [1]: Explain shortly the following terms. (6 points)

- (a) Genetic map-function.
- (b) Penetrance probability.
- (c) Non-parametric linkage analysis.
- (d) Genetic heterogeneity.
- (e) Linkage disequilibrium (LD).
- (f) Hardy-Weinberg equilibrium (HWE).

Question [2]: Formulate the mixed model (= genetic term) which is also sometimes called mixed inheritance model. Explain the terms (effects) in the model and state the typical assumptions. (6 points)

Question [3]: Explain with your own words what are the basic ideas in linkage analysis and in association analysis. Try to explain what differences there are. Statistical details can be kept to a minimum. (6 points)

Question [4]: Let us have a pedigree data and the markers (M_1, M_2, M_3, M_4, M_5) with recombination fractions ($\theta_1, \theta_2, \theta_3, \theta_4$) between markers. When we place the trait locus l on the position between the markers M_3 and M_4 , we obtain recombination fractions θ_L (between M_3 and the trait locus l) and θ_R (between trait locus l and M_4). Please explain then shortly how can we calculate the multipoint likelihood (for these markers and the trait locus l jointly) using the Lander-Green algorithm. (6 points)

Question [5]: Outline with your own words the basic ideas in Haseman-Elston sib-pair analysis for quantitative traits. Explain also terms IBD and IBS here. (6 points)