EXAM / 20. Oct 2010 / C128 Computer class

Send answers to sirkka-liisa.varvio@helsinki.fi, use **.doc, .txt or .pdf.**

Include your student number.

Before leaving the classroom: check from Siru that your e-mail has been received.

You get max 30 points from this exam and 20 points from returned assignments (answers need not be correct !). 25 points -> grade 1, 30 points -> grade 2, 35 points -> grade 3, 40 points -> grade 4, 45-50 points grade 5. You´ll get correct answers and grade & credits registration to weboodi within a week. Later you can extend you credits by extra assignments if you like. Extra assignments will be available during next 3 weeks.

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1. (16 points)

Exam201011 dataset includes 30 sequence items.

(a) (3 points) From what organism(s) are the sequences?
(b) (3 points) What is name of the gene from which the sequences are?
(c) (5 points) Construct neighbor-joining, UPGMA and parsimony trees from the data. You can use any model of nucleotide substitution (i.e. you don´t have to pay attention to model selection.)
(d) (5 points) Explain what you see in the resulting trees and why do they look different.

2. (4 points)

In the Hardy-Weinberg "equilibrium" the allele frequencies and genotypes frequencies have a certain relationship (Two alleles with frequencies p and q, three genotypes with frequencies p^2 and $2pq$ and q^2 ($p + q = 1$ and $p^2 + 2pq + q^2 = 1$). What about a case in which there are three alleles, $p + q + r = 1$. What are the Hardy-Weinberg genotype frequencies?

Human ABO blood group system has three major alleles (in addition more than 70 molecularly distinct alleles): I^A, I^B and I^O. Allele I^O is recessive as regards alleles I^A and I^B (I^O gene has a single nucleotide deletion which results in inactive protein). Genotypes I^AI^A and I^AI^O are phenotypically A (the blood group A), I^BI^B and I^BI^O are phenotypically B (the blood group B), I^AI^B is phenotypically AB and $I^O I^O$ is O (the blood group O). Allele frequencies in one human population are: 0.3 for I^A, 0.6 for I^B and 0.1 for I^O. Assuming Hardy-Weinberg equilibrium, what are the blood group phenotype frequencies in the population?

3. (4 points)

Four OTU sequences are the material for maximum parsimony and for maximum likelihood phylogeny inference analysis. Focus is in one particular nucleotide site (as in the examples in lecture slides). Why is it that many more possibilities must be considered for maximum likelihood procedure than for parsimony procedure?

4. (4 points)

A disease is caused by a recessive allele with simple Mendelian inheritance. Mary has a sister with this disease and Bill has a brother with this disease. There are no other known cases in their families. What is the probability that Mary´s and Bill´s first child is a girl who will have the disease?

5. (2 points)

Sickle cell anemia is a famous example about balancing selection and a balanced polymorphism in geographical regions where malaria exists. In a certain region malaria acts so that 30% humans without the sickle cell allele die because of malaria and 20% of humans homozygous for the sickle cell allele die because of anemia. What are the frequencies of the normal allele and sickle cell allele in an equilibrium?