Human Genetics:
Problem Set VIII

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Abstract

This work contains the solutions to the problem set VIII of Human Genetics 2015 course at New York University.

Question 1.

Solution. (a) The variant site is not located in a transcribed region of the lactase gene, but still can cause lactose intolerance, as we saw in class that the labeled fragments can be arbitrarily long in its size.

(b) Under the assumption of Hardy-Weinberg principle, we have

$$\begin{aligned} p(TT) &= p^2 \\ &= 0.71^2 \approx 0.504 \\ p(CT) &= 2pq \\ &= 2(0.71)(1-0.71) \approx 0.412 \\ P(CC) &= qq \\ &= (1-0.71)^2 \approx 0.0841, \end{aligned}$$

for the expected frequency of each of the three genotype classes at the locus among Swedes.

(c) We know that TT and CT genotypes yield the lactose-tolerant phenotype in an individual. Therefore, the proportion of Swedish adults who can eat their herring with cream cheese is $p(TT) + p(CT) \approx 0.504 + 0.412 \approx 0.916$.

□

Question 2.

Solution. (a) 5'-CACUAA... $\rightarrow$ 5'-CACUAG... would produce a nonsense mutation, as the UAA base changed to UAG, making amino acid change to STOP.

(b) 5'-CAC... $\rightarrow$ 5'-CAU... would produce a silent mutation, as the CAC base and CAU base have the same amino acid, His.

(c) 5'-CAC... $\rightarrow$ 5'-CCC would produce a missense mutation, as the amino acid changes from His to Pro. □

Question 3.

Solution. (a) At Hardy-Weinberg equilibrium, the expected frequency of color-blindness among women is q^2, where q denotes the frequency of mutant allele in the population, which in this case is $0.1^2 = 0.01$.

(b) The expected frequency for men is simply $q = 0.1$ as, an color-blind man will be hemizygous, requiring exactly one mutant allele.

□