Biol3120 Problem set 3

1. You are studying a population in Hardy-Weinberg equilibrium, where eye colour is controlled by a single gene. This gene has 3 alleles:

A1 is dominant to A2 and A3, and the phenotype is brown eyes

A2 is dominant to A3, and the phenotype is blue eyes

A3 phenotype is green eyes.

a) What eye colour would be expected for someone whose genotype is A2A3?

a) Green
b) Blue
c) Brown

b) If the frequency of the A1 allele is 0.50, and the frequency of the A2 allele is 0.30, what is the frequency of people in this population who are brown-eyed, but carry the green eye allele?

2. The prevalence of Sandhoff disease, an autosomal recessive condition, is 5 per million people. **Assuming that there is no selection at this gene locus and that mating is random within the population, how many of Australia's 25.69 million people would you expect to be carriers for the condition?**

3. A woman is affected with Alpha-1 antitrypsin deficiency, an autosomal recessive condition with a carrier frequency of 1 in 16. As they are planning on having children, her partner undergoes carrier testing for the condition. The result was negative, indicating that he was not found to be a carrier. The reported sensitivity of the carrier testing is 95%.
What is the chance that their first child will be affected with Alpha-1 antitrypsin deficiency?

4. The Huntington's Disease (HD) gene (HTT) is located on chromosome 4, and the age of onset of the condition is closely related to the repeat-expansion size within this gene.
Recent research shows that a particular variant on chromosome 15 acts as a modifier for the condition. Someone with a repeat expansion in their HTT gene as well as this modifier variant will develop HD symptoms approximately six years earlier than someone without this modifier variant.
A woman with Huntington's disease has one copy of this modifier variant, and has a child with an unaffected man, who also has one copy of this modifier variant.

a) What are the chances their child has inherited the HD expansion, but not the modifier variant?

b) What are the chances their child has inherited the HD expansion and at least one copy of the modifier variant?

5. A mutation is detected in a gene: at basepair 735 of the mRNA transcript a T has changed to a C. The coding region of the gene starts after basepair 420 of the mRNA transcript.
   Hint: The DNA change is described in terms of the coding region, not the position on the mRNA transcript.
   Write the DNA mutation in standard naming format:

6. Name genetic variants
   A mutation is detected in a gene: between basepairs 126 and 127 of the coding region, a 3 basepair insertion of TAA has occurred.
   Write the DNA mutation in standard naming format:

7. **What impact might this have on the expressed gene?**

8. **Which set of chromosomal structural changes are most likely to result in a copy number variant?**
   a. Duplication and Inversion
   b. Duplication and Deletion
   c. Duplication and Insertion
   d. Deletion and Inversion