1. Hemophilia is caused by an X-linked recessive allele. In the pedigree shown below which two individuals in the pedigree must be carriers of hemophilia?

- A. I-1 and II-1
- B. I-4 and II-2
- C. II-1 and II-2
- D. III-2 and III-3

2. A single gene in humans causes blood to be either rhesus positive (dominant allele) or rhesus negative (recessive allele). A woman with rhesus negative blood has already had a child with rhesus positive blood. There could be complications during pregnancy if she has another child with rhesus positive blood. What is the probability of this, if the father is the same, and if his mother is known to have rhesus negative blood?

- A. 25%
- B. 50%
- C. 75%
- D. 100%

3. What does the genotype \(X^H X^h\) indicate?

- A. A co-dominant female
- B. A heterozygous male
- C. A heterozygous female
- D. A co-dominant male

4. (a) Define the term sex linkage.

(b) A male and female with normal colour vision each have a father who is colour blind. They are planning to have children. Predict, showing your working, the possible phenotypes and genotypes of male and female children.
5.  (a) Define the term **co-dominance**.

When both alleles are expressed in the phenotype.

(b) A man of blood type AB and a woman of blood type B are expecting a baby. The woman’s mother had blood type O. Deduce the possible phenotypes of the offspring from the cross shown below.

Parent phenotypes  

<table>
<thead>
<tr>
<th></th>
<th>AB</th>
<th>B</th>
</tr>
</thead>
</table>

Parents genotypes  

|   | IA | IB | I^B | I^b |

Parent gametes  

|   | IA | IB | I^B | I^b |

|   | 0^a | IA | IB |

|   | 1^b | IA | I^B | I^b |

|   | IA | I^b | IB | I^B |

F\(_1\) genotypes  

|   | IA| IB| I^B| I^b |

F\(_1\) phenotypes  

|   | AB blood | A blood | B blood | B blood |

1/4 AB blood, 1/4 A blood, 1/2 B blood

6.  (a) What is the difference between an allele, a gene, a genome?

- **Allele** - a form of a gene
- **Gene** - a heritable factor that consists of a length of DNA and influences a specific characteristic
- **Genome** - the whole genetic information of an organism

(b) What was the Human Genome Project?  

A project undertaken to sequence the entire genome of a human. There are 23,000 protein coding genes. Most of the genome is considered "junk DNA", (that affect gene expression as well as areas of highly repetitive sequences called satellite DNA)
7. (a) Compare and contrast sickle cell anemia to cystic fibrosis.

- Sickle cell anemia
- Caused by codominant alleles
- Results in red blood cells are distorted into a sickle shape
- Neither disorder is sex linked

- Cystic Fibrosis
- Recessive alleles
- Non-functional chloride channels causing mucus buildup in lungs and blockages in pancreatic ducts
- Huntington's disease
- Dominant allele on chromosome 4
- Brain degeneration leading to changes in behavior, thinking, emotions

(b) Compare and contrast color blindness to Huntington’s disease.

- Color blindness
- Caused by recessive allele on X chromosome
- Results in cannot clearly distinguish certain colors (such as red-green)

- Both disorders result in producing a faulty protein

8. A gene in humans called APC is located on chromosome 5. This gene controls cell division and is known as a tumour suppressor gene. Mutations of APC cause a genetic disease called FAP (Familial Adenomatous Polyposis).

(a) State, with a reason, whether FAP is a sex-linked genetic disease or not.

Not sex linked genetic disease because gene is not on a sex chromosome

(1) 50% of the gametes produced by a person with FAP have an APC gene with the mutation.

(b) Identify, with a reason, whether FAP follows a dominant or recessive pattern of inheritance.

Dominant

Person with FAP has one mutant and one normal allele of the gene if heterozygous

If recessive 100% of the gametes would have the mutated APC gene

(2)
In a person with FAP, each cell contains a copy of the APC gene with the mutation. If a mutation occurs on the cell’s other copy of the APC gene, the cell becomes a tumour cell. Almost everyone with FAP develops cancer before the age of 50.

(c) Explain why almost everyone with FAP eventually develops cancer.

- only one cell needs to have the mutation to its APC gene (mutation has to occur only once)
- the tumor develops from one cell
- all the cells of the body could become tumour cells
- if both alleles for the APC gene are mutated the tumor still develops