1: pedigrees, genetic mapping, genome organization.

Question	Marks	Question type
i		Information or resources
1.1	1	Multiple Choice
1.2	1	Multiple Choice
1.3	1	Multiple Choice
1.4	1	Multiple Choice
1.5	1	Multiple Response
1.6	1	Multiple Choice
1.7	2	Essay
1.8	2	Essay
1.9	2	Essay
1.10	2	Essay
1.11	1	Essay

2: genetic variation and testing

Question	Marks	Question type
2.1	1	Multiple Choice
2.2	1	Multiple Choice
2.3	1	Multiple Choice
2.4	1	Multiple Choice
2.5	1	Multiple Choice

2.6	1	Multiple Choice
2.7	2	Essay
2.8	1	Essay
2.9	4	Essay
2.10	2	Essay

3: gene expression

Question	Marks	Question type
3.1	1	Multiple Choice
3.2	1	Multiple Response
3.3	1	Multiple Choice
3.4	1	Multiple Choice
3.5	2	Essay
3.6	2	Essay
3.7	2	Essay

1.7 Describe what is a long non-coding RNA (lncRNA), and give two examples of functions (2p)
Skriv ditt svar här. Ändringar sparas automatiskt.

Format

Words: 0

Maximum marks: 2

1.8 Briefly explain autozygosity mapping, and in what type of families this is a good method to find disease causing genes. (2p)

Skriv ditt svar här. Ändringar sparas automatiskt.

Format

Words: 0

Maximum marks: 2

1.9 We want through linkage analysis localize the gene causing a dominantly inherited disease. We have DNA from a family, and got the following result for three different polymorphic markers (D, E and F) on chromosome 5.

Paternal grandmother (Disease): Marker **D: 1, 2** ; Marker **E: 1, 2** ; Marker **F: 3, 4**

Paternal grandfather (healthy): **D**: **3, 3** ; **E**: **1, 1** ; **F**: **1, 4**

Father (Disease): **D**: **1, 3** ; **E**: **1, 2** ; **F**: **4, 4**

Mother (healthy): **D**: **3, 4** ; **E**: **2, 2** ; **F**: **1, 3**

Daughter 1 (healthy): **D**: **1, 3** ; **E**: **1, 2** ; **F**: **1, 4**

Son (healthy): **D**: **1, 4** ; **E**: **1, 2** ; **F**: **3, 4**

Daughter 2 (Disease): **D**: **3, 4** ; **E**: **2, 2** ; **F**: **1, 4**

a) What marker/markers are linked to the disease (1p)?

b) What marker gives the highest lod score (1p)?

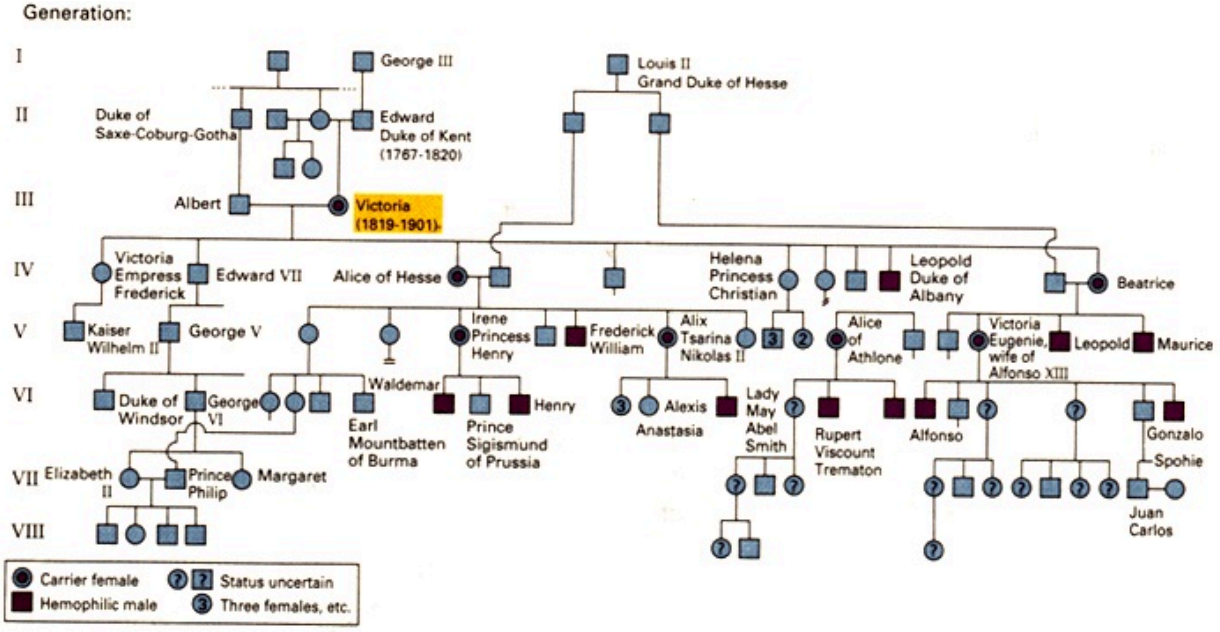
Fill in your answer here

Format

Words: 0

Maximum marks: 2

1.10 Haemophilia has been present in the British royal family for centuries, with queen Victoria (1819-1901) being the first known carrier in the lineage. The pedigree bellow shows the carrier/disease status of many members of the Victorian Dynasty. Explain how the disease is inherited and why women are mostly carriers (1p). Based on the pedigree, identify one piece of evidence that strongly supports your identified mode of inheritance in this family, and explain why it supports this mode of inheritance (1p). (2p)



Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 2

1.11 With reference to the pedigree below explain how reduced penetrance or variable expressivity could make a dominant disorder appear to “skip” generations. (1p)

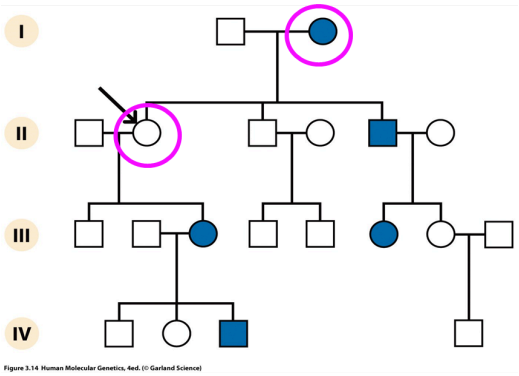


Figure 3.14 Human Molecular Genetics, 4ed. (© Garland Science)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 1

2.7 Explain why C-T transition is the most frequent single nucleotide change in our DNA. (2p)
Skriv ditt svar här. Ändringar sparas automatiskt.

Format

Words: 0

Maximum marks: 2

2.8 Give one example of a structural variant that can be found in the human genome, and briefly explain how it looks like. (1p)

Skriv ditt svar här. Ändringar sparas automatiskt.

Format

Words: 0

Maximum marks: 1

2.9 Genetic testing in clinical practice requires choosing the correct testing strategy and understanding both technical and interpretational limitations. Using examples from the lecture, discuss:

-How different genetic testing methods (single-gene testing, gene panels, chromosomal microarray, WES/WGS) vary in their diagnostic utility and appropriate clinical indications. (2p)

-The main technical limitations of next-generation sequencing (NGS), including variant types that are difficult or impossible to detect. (1p)

-How variant interpretation challenges—such as penetrance, VUS, and population allele frequency—affect the clinical usefulness of genetic test results. (1p) (Total 4p)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 4

2.10 Array-based comparative genome hybridization (aCGH) are sometimes used to investigate unexplained developmental delay. Explain how aCGH work (1p), and what kinds of genetic changes it detects (1p). (Total 2p)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 2

3.5 Explain why the phenotype is determined by gene regulation rather than gene sequence alone (2p)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 2

3.6 Explain the post transcriptional RNA processing 5’ capping and polyadenylation (1p), and explain their function in mRNA maturation (1p). (Total 2p)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 2

3.7 Briefly explain what *ten-eleven translocation (TET) enzymes* do to methylated cytosines (1p) and how this allows methylation to be reversed (1p). (Total 2p)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 2