























1. What kind of inheritance is represented by the following pedigree chart?



1. sex-linked
2. co-dominant
3. autosomal recessive
4. autosomal dominant
5. A forensic officer collected biological evidence that was lacking nucleated cells from a crime scene. What biological technique may the investigator undertake to help detectives find the perpetrator?
6. DNA analysis
7. Marker analysis
8. PCR analysis
9. mtDNA analysis
10. The ABO blood system is an example of which type of genetic inheritance?
11. Multiple Allele Inheritance
12. Sex-linked Inheritance
13. Simple Inheritance
14. Polygenic Inheritance



1. The type of inheritance displayed in the pedigree chart is best described as
	1. autosomal dominant.
	2. autosomal recessive.
	3. co-dominant.
	4. sex-linked.

Huntington’s disease (HD) is an inherited neurodegenerative disorder due to a defective gene located on chromosome 4. Parents-to-be with a history of HD in their families often visit a genetic counsellor to discuss the possibilities and implications of the disease.

1. It is found that a husband is heterozygous for HD whilst the wife is homozygous recessive.

In the space below, predict the possible genotypes and phenotypes of their future children.

(5 marks)

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1. Construct a pedigree diagram the genetic counsellor may have drawn to explain the couple’s chances of inheritance.

Your pedigree should include:

* two generations of the family
* the wife’s sister and brother whom are all unaffected
* the husband’s older brother who died from HD symptoms
* the prediction of two future children, one of each gender

(7 marks)

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1. Describe a suitable prenatal genetic test that could be used to determine if an unborn child has HD, and state one risk associated with the test.

(3 marks)

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The subjects of the study were required to have samples of their DNA taken for profiling by scientists.

1. State the name and outline the process used to create DNA fingerprints.

(6 marks)

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1. Discuss ethical implications that should be considered when using genetic profiling.

(4 marks)

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1. Individual III-3 bred with a female carrier and gave birth to a daughter. What is the probability of the daughter inheriting this trait?
2. 0%
3. 25%
4. 50%
5. 75%
6. An example of an X-linked recessive disease would be
7. Haemophilia.
8. Huntington’s disease.
9. Phenylketonuria (PKU).
10. Fragile X Syndrome.
11. Describe how ABO blood groups are inherited. (3 marks)

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1. Using a Punnett square, explain if it is possible for an O-type child to be born to a B-type father and AB-type mother. (4 marks)

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**Question 33 (14 marks)**

Shown below is a pedigree that outlines the inheritance of a particular disease within a family.

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(a) Using evidence from the pedigree, explain why the disorder cannot be transmitted by a sex-linked recessive gene. (2 marks)

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(b) Using evidence from the pedigree, explain why the disorder cannot be transmitted by an autosomal dominant gene. (2 marks)

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(c) Assume that the individual II 1 has a genotype **Bb**. Individuals II 1 and II 2 have another child. What is the probability that the child will be an affected daughter? Show your working. (4 marks)

(d) Which male in generation 3 is more **unlikely** to pass on the allele to his future children? Explain your reasoning. (3 marks)

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(e) If ll5 becomes pregnant again, describe **one** prenatal test this couple could have to find out the genotype of the unborn child and state **one** risk associated with this method of prenatal testing. (3 marks)

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**Question 34 (11 marks)**

Paternity testing involves a number of procedures used to determine the biological father of a child. One of these is testing for blood groups while the other looks at DNA profiling.

The following shows some results for these two procedures.

**Procedure 1: Testing for ABO blood groups**

Mother’s blood group: B

Child’s blood group: O

Possible biological father’s blood groups – Father 1: O, Father 2: A, Father 3: B

**Procedure 2: DNA profile data**



(a) (i) From the ABO blood group data, identify which male(s), can be

 excluded as the possible biological father of the child. (1 mark)

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(ii) Using Punnett squares, show how you arrived at your answer in part (a)(i). (4 marks)

(b) Describe the process that produces DNA profile data. (4 marks)

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(c) Using the data from the DNA profile, suggest which of the fathers is the actual father of the child. Explain how you arrived at your answer. (2 marks)

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Questions 17,18 and 19 refer to the diagram below, which shows the inheritance of a disorder within a family.

 

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 3

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 1

 2

 1

 III

 II

 I

1. The mode of inheritance depicted above is
	1. autosomal recessive.
	2. sex-linked recessive.
	3. autosomal dominant.
	4. sex-linked dominant.

18. The **best** evidence from this pedigree that depicts the mode of inheritance is

* 1. an affected parent has an unaffected child.
	2. the trait appears in males and females in equal numbers.
	3. the trait skips generations.
	4. two unaffected parents have an affected offspring.

19. If individuals II1 and II2 were to have another child, what would be the chance of the

 child being affected by this disorder?

* 1. 100%
	2. 75%
	3. 50%
	4. 25%

**Question 34 (9 marks)**

The inheritance of genes located on the sex chromosomes produces a phenotype ratio that is different to those that are obtained when looking at inheritance of genes found on autosomes. Inheritance of genes that are found on the X chromosome is referred to as X-linked inheritance.

1. State the sex chromosome configuration for males. (1 mark)

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1. Colour blindness is an example of X-linked recessive inheritance. Using the letters **B** and **b**, determine the genotypes of the following individuals. (3 marks)

(i) Colour-blind male \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

(ii) Unaffected female \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

(iii) Unaffected male \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

 (c) Describe why a father with an X-linked disorder cannot pass the condition onto his sons. (2 marks)

 \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

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 (d) Explain how the process of amniocentesis can be used to detect genetic disorders.

 (3 marks)

 \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

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1. What kind of inheritance is represented by the following pedigree chart?



1. sex-linked
2. co-dominant
3. autosomal recessive
4. autosomal dominant
5. A forensic officer collected biological evidence from a crime scene that was lacking nucleated cells. What biological technique may the investigator undertake to help detectives find the perpetrator?
6. DNA analysis
7. Marker analysis
8. PCR analysis
9. **mtDNA analysis**
10. The ABO blood system is an example of which type of genetic inheritance?
11. **Multiple Allele Inheritance**
12. Sex-linked Inheritance
13. Simple Inheritance
14. Polygenic Inheritance

**Use the pedigree chart below to answer Question 29.**



1. The type of inheritance displayed in the pedigree chart is best described as
	1. autosomal dominant.
	2. **autosomal recessive.**
	3. co-dominant.
	4. sex-linked.

Huntington’s disease (HD) is an inherited neurodegenerative disorder due to a defective gene located on chromosome 4. Parents-to-be with a history of HD in their families often visit a genetic counsellor to discuss the possibilities and implications of the disease.

1. It is found that a husband is heterozygous for HD whilst the wife is homozygous recessive.

In the space below, predict the possible genotypes and phenotypes of their future children.

|  |  |
| --- | --- |
|  | **Mother** |
| **h** | **h** |
| **Father** | **H** | **Hh** | **Hh** |
| **h** | **hh** | **hh** |

(5 marks)

|  |  |
| --- | --- |
| **Punnett Square:****Father’s genes: Hh (heterozygous)****Mother’s gene: hh (homozygous)** | **2** |
| **Key: H – Huntington’s gene h – Normal gene** | **1** |
| **Phenotype: 50% Huntington’s and 50% Normal** | **1** |
| **Genotype: 50% Heterozygous, 50% homozygous normal/recessive** | **1** |

1. Construct a pedigree diagram the genetic counsellor may have drawn to explain the couple’s chances of inheritance.

(7 marks)

Your pedigree should include:

* two generations of the family
* the wife’s sister and brother whom are all unaffected
* the husband’s older brother who died from HD symptoms
* the prediction of two future children, one of each gender

|  |  |
| --- | --- |
| **Use of a key** | **1** |
| **Couple and their siblings in generation 1** | **1** |
| **Children in generation 2** | **1** |
| **Husband’s brother with HD** | **1** |
| **Wife and siblings not affected** | **1** |
| **Possible offspring (one male, one female)** | **1** |
| **50% offspring with HD** | **1** |



**Note: Either child may be represented as having HD**

1. Describe a suitable prenatal genetic test that could be used to determine if an unborn child has HD, and state one risk associated with the test.

(3 marks)

**Any ONE of the following:**

|  |  |
| --- | --- |
| **Description** | **Marks** |
| Chorionic Villus Sampling | 1 |
| Obtain foetal cells from chorion membrane for analysis | 1 |
| Risk of miscarriage | 1 |

|  |  |
| --- | --- |
| **Description** | **Marks** |
| Amniocentesis | 1 |
| Obtain sample of amniotic fluid cells for analysis | 1 |
| Risk of infection / miscarriage / damage to the foetus | 1 |

|  |  |
| --- | --- |
| **Description** | **Marks** |
| Foetal Blood Sampling | 1 |
| Obtain blood samples from the placenta | 1 |
| Risk of infection / bleeding from sample site / leaking of amniotic fluid / miscarriage / change in foetus heart rate | 1 |

The subjects of the study were required to have samples of their DNA taken for profiling by scientists.

1. State the name and outline the process used to create DNA fingerprints.

(6 marks)

|  |  |
| --- | --- |
| **Description** | **Marks** |
| (Gel) Electrophoresis  | 1 |
| DNA cut by restriction enzymesSections of DNA / DNA pieces placed at one end of gel bed Electric current is passed through the gel / voltage applied Negatively charged DNA moves towards positive electrode / terminalDNA moves through the gel at different speeds / smaller moves faster than largerBands form representing different size fragments of DNA | 1-5 |

1. Discuss ethical implications that should be considered when using genetic profiling.

(4 marks)

**Any four of the following or other suitable answers:**

* **Who the genome/profile belongs to / Ownership of the genome/profile**
* **Privacy/confidentiality of the subject**
* **Potential for discrimination**
* **Inappropriate applications of genetic testing**
* **Misuse of genome/profiles such as DNA data banks**

**Questions 17 and 18 refer to the X-linked recessive pedigree below.**

****

1. Individual III-3 bred with a female carrier and gave birth to a daughter. What is the probability of the daughter inheriting this trait?
2. 0%
3. 25%
4. **50%**
5. 75%
6. An example of an X-linked recessive disease would be
7. **Haemophilia.**
8. Huntington’s disease.
9. Phenylketonuria (PKU).
10. Fragile X Syndrome.
11. Describe how ABO blood groups are inherited. (3 marks)

|  |  |
| --- | --- |
| **Description** | **Mark** |
| Any three of the following for one mark each: |  |
| * Multiple allele inheritance
 | 1-3 |
| * ABO gene is autosomal
 |
| * AB groups are dominant over O / O is recessive
 |
| * AB groups are co-dominant
 |
| **Total** | **3** |

1. Using a Punnett square, explain if it is possible for an O-type child to be born to a B-type father and AB-type mother. (4 marks)

|  |  |
| --- | --- |
| **Description** | **Mark** |
| Identifies mother can only be AB/IAIB | 1 |
| Identifies father could be BO/IBi or BB/IBIB | 1 |
| Correct Punnett square/s used | 1 |
| No possibility of O-type child to be born | 1 |
| **Total** | **4** |

**Question 33 (14 marks)**

Shown below is a pedigree that outlines the inheritance of a particular disease within a family.

****

(a) Using evidence from the pedigree, explain why the disorder cannot be transmitted by a sex-linked recessive gene. (2 marks)

|  |  |
| --- | --- |
| **Description** | **Mark** |
| If sex linked then an affected female such as I1 would pass it on to her son II4, but he is not affected | 1 |
| III7 is affected but II6 is not | 1 |
| **Total** | **2** |

(b) Using evidence from the pedigree, explain why the disorder cannot be transmitted by an autosomal dominant gene. (2 marks)

|  |  |
| --- | --- |
| **Description** | **Mark** |
| If it was autosomal dominant then two unaffected parents could not have an affected child  | 1 |
| II 5 and II6 are unaffected which means they do not have the dominant allele but they have produced an affected offspring III7 | 1 |
| **Total** | **2** |

(c) Assume that the individual II 1 has a genotype **Bb**. Individuals II 1 and II 2 have another child.

 What is the probability that it will be an affected daughter? Show your working.

 (4 marks)

 PI Bb x bb

 B b

|  |  |
| --- | --- |
| Bb | bb |
| Bb | bb |

b

b

2 Bb: 2 bb

|  |  |
| --- | --- |
| **Description** | **Mark** |
| * Correct genotypes of parents listed
* Correct genotypes of potential offspring inside Punnett square
 | 11 |
| Probability of producing a daughter with disease = ½ x ½ = ¼ or 0.25 | 11 |
| **Total** | **4** |

(d) Which male in generation 3 is more **unlikely** to pass on the allele to his future children? Explain your reasoning. (3 marks)

|  |  |
| --- | --- |
| **Description** | **Mark** |
| III5 | 1 |
| II3 must have one b allele as they have an affected parent therefore the probability of passing it on to his children is 50% | 1 |
| III5 could be BB or Bb therefore the chance of passing the b on to his children lower/less than III3 | 1 |
| **Total** | **3** |

 (e) If ll5 becomes pregnant again describe **one** prenatal test this couple could have to find out the genotype of the unborn child and state **one** risk associated with this method of prenatal testing. (3 marks)

|  |  |
| --- | --- |
| **Description** | **Mark** |
| **Any one of the following examples.**  |  |
| **1. Amniocentesis**Removal of amniotic fluid (16th-20th week pregnancy) from amniotic sac – about 10 – 20 mlFoetal cells in fluid examined | 1-2 |
| Risk- infection, miscarriage or damage to baby | 1 |
| **2. Chorionic villus sampling**Obtain foetal cells from chorion using needle aspiration.Cells examined in the same way as amniocentesis | 1-2 |
| Risk- miscarriage | 1 |
| **3. Foetal blood sampling**obtain foetal blood samples from placenta | 1-2 |
| Risk of infection / bleeding from sample site / leaking of amniotic fluid / miscarriage / change in foetus heart rate | 1 |
| **Total** | **3** |

**Question 34 (11 marks)**

Paternity testing involves a number of procedures used to determine the biological father of a child. One of these is testing for blood groups while the other looks at DNA profiling.

The following shows the results of these two procedures.

**Procedure 1: Testing for ABO blood groups**

Mother’s blood group: B

Childs blood group: O

Possible biological father’s blood groups – Father 1: O, Father 2: A, Father 3: B

**Procedure 2: DNA profile data**



(a) (i) From the ABO blood group data, identify which male(s), can be excluded as the possible biological father of the child. (1 mark)

|  |  |
| --- | --- |
| **Description** | **Mark** |
| None | 1 |
| **Total** | **1** |

(ii) Using Punnett squares show how you arrived at your answer in part (a)(i). (4 marks)

 **Possible father 1**

 Father

Mother

|  |  |  |
| --- | --- | --- |
|  | i | i |
| IB | IB i | IB i |
| i | ii | ii |

 **Possible father 2**

 Father

Mother

|  |  |  |
| --- | --- | --- |
|  | IA | i |
| IB | IAIB | IBi |
| i | IAi | ii |

 **Possible father 3**

 Father

Mother

|  |  |  |
| --- | --- | --- |
|  | IB | i |
| IB | IBIB | IBi |
| i | IBi | ii |

|  |  |
| --- | --- |
| **Description** | **Mark** |
| 1 mark per Punnett square | 3 |
| All fathers could produce an O blood group child  | 1 |
| **Total** | **4** |

(b) Describe the process that produces DNA profile data.

 (4 marks)

|  |  |
| --- | --- |
| **Description** | **Marks** |
| **Any 4 of the following** |  |
| * DNA cut by restriction enzymes
* Sections of DNA / DNA pieces placed at one end of gel bed
* Buffer solution bathes gel
* Electric current is passed through the gel / voltage applied
* Negatively charged DNA moves towards positive electrode / terminal
* DNA moves through the gel at different speeds / smaller moves faster than larger
* Bands form representing different size fragments of DNA
 |  1-4 |
| **Total** | **4** |

 (c) Using the data from the DNA profile suggest which of the fathers is the actual father of the child. Explain how you arrived at your answer. (2 marks)

|  |  |
| --- | --- |
| **Description** | **Mark** |
| Possible father 1 | 1 |
| Only father 1 has matching bands in places where mother’s bands do not match those of the child. | 1 |
| **Total** | **2** |

|  |  |
| --- | --- |
| 17 | A |
| 18 | D |
| 19 | D |

**Question 34 (9 marks)**

1. What is the sex chromosome configuration for males. (1 mark)

|  |  |
| --- | --- |
| **Description** | **Mark** |
| males : XY | 1 |
| **Total** | **1** |

1. Colour blindness is an example of x-linked recessive inheritance. Using the letters **B** and **b**, determine the genotypes of the following individuals. (3 marks)

(i) Colour-blind male \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

(ii) Unaffected female \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

(iii) Unaffected male \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

|  |  |
| --- | --- |
| **Description** | **Mark** |
| (i) XbY | 1 |
| (ii) XBXB or XBXb (must have both for mark) | 1 |
| (iii) XBY | 1 |
| **Total** | **3** |

 (c) Describe why a father with an X-linked disorder cannot pass the condition onto his son (2 marks)

|  |  |
| --- | --- |
| **Description** | **Mark** |
| Y chromosome does not carry colour blindness gene | 1 |
| Sons can only inherit Y gene from father as this is what makes them male. | 1 |
| **Total** | **2** |

 (d) Explain how the process of amniocentesis can be used to detect genetic disorders.

 (3 marks)

|  |  |
| --- | --- |
| **Description** | **Mark** |
| Removal of amniotic fluid (16th-20th week pregnancy) | 1 |
| Foetal cells found in amniotic fluid | 1 |
| Cells examined for biochemical defects/ chromosomal abnormalities | 1 |
| **Total** | **3** |