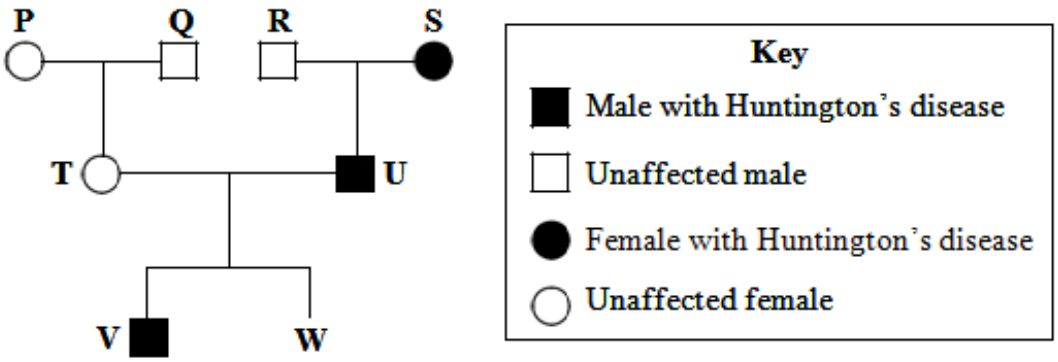
Inherited Disorders 2

Q:1 Huntington's disease is a disorder of the nervous system. It is caused by a dominant allele. The family tree shows the inheritance of Huntington's disease in one family.



(a) T and U already have a son, V, with Huntington's disease. What is the chance that their next child, W, will also develop Huntington's disease?

Use a genetic diagram in your answer.

Use the symbols H to represent the dominant allele and h to represent the recessive allele.

Chance that child W will have Huntington's disease = ...

(4 marks)

(b) A doctor advises parents T and U that embryo W should be 'screened'.

(b)(i) What is embryo screening?

_____

_____

_____

(1 mark)

(b)(ii) Some people do not believe that embryos should be screened.

Give one reason why.

_____

_____

_____

(1 mark)

(c) Cystic fibrosis is an inherited disorder in which the person's mucus becomes thick.

(c)(i) For this gene, one allele in every 30 in the population is the cystic fibrosis allele.

However, only one person in every 900 has cystic fibrosis.

Explain why.

_____

_____

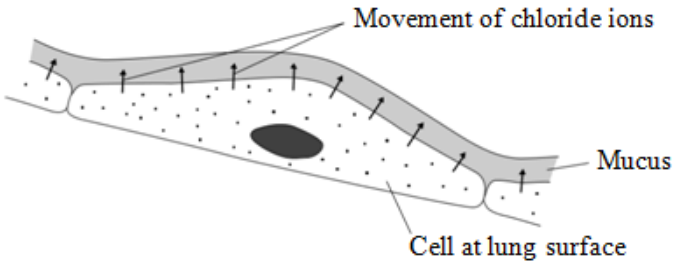
_____

_____

_____

(2 marks)

(c) (ii) The diagram shows how, in a healthy person, cells at the lung surface move chloride ions into the mucus surrounding the air passages.



This movement of chloride ions causes water to pass out of the cells into the mucus.

Explain why.

_____

_____

_____

_____

_____

_____

_____

(3 marks)

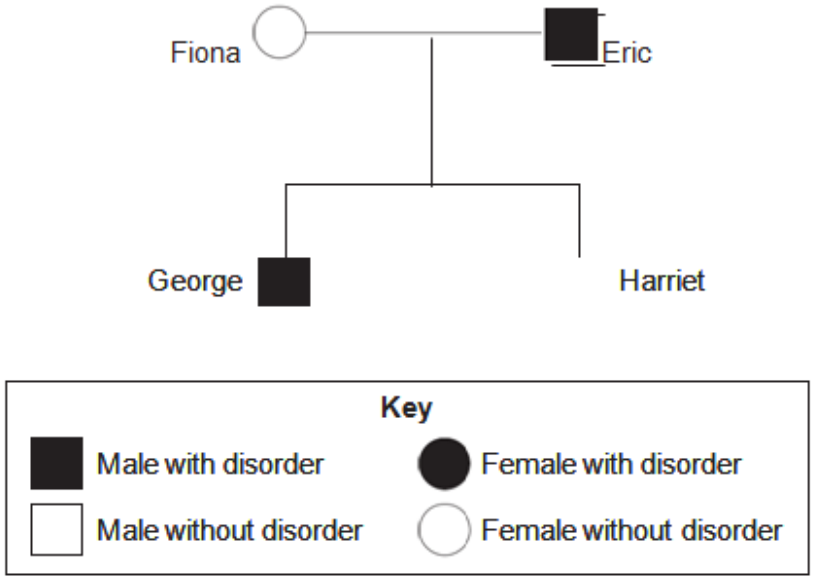
(c) (iii) Mucus contains protein.

Which parts of the lung cells manufacture the protein in mucus?

_____

(1 mark)

Q:2 The family tree shows the inheritance of a disorder caused by a dominant allele. Fiona and Eric have two children George and Harriet.



(a) The son, George, has the disorder.

The daughter, Harriet, does not have the disorder.

(a) (i) Use the key to draw the symbol for Harriet next to her name on the family tree.

(2 marks)

(a) (ii) The symbol D represents the dominant allele for the disorder.

The symbol d represents the recessive allele.

Fiona has the pair of alleles dd.

Write the correct pairs of alleles in the boxes.

Harriet has the pair of alleles ☐ .

A person with the disorder could have

the pair of alleles ☐ or the pair of alleles . ☐

(3 marks)

(b) Before Harriet was born, a doctor suggested that Fiona should have the embryo 'screened'.

(b) (i) Give one reason why the doctor suggested screening.

Tick (☒) one box.

To check for the D allele ☐

To check the sex of the embryo ☐

To cure the disorder ☐

(1 mark)

(b) (ii) Why do some people believe that embryos should not be screened?

_____

_____

(1 mark)

Q:3 Cystic fibrosis is an inherited disorder.

Mr and Mrs Brown do not have cystic fibrosis but they have a child with cystic fibrosis.

(a) Draw a ring around the correct answer to complete each sentence.

(a) (i) The allele for cystic fibrosis is a

carrier allele.

dominant allele.

recessive allele.

(1 mark)

(a) (ii) Mr and Mrs Brown are both

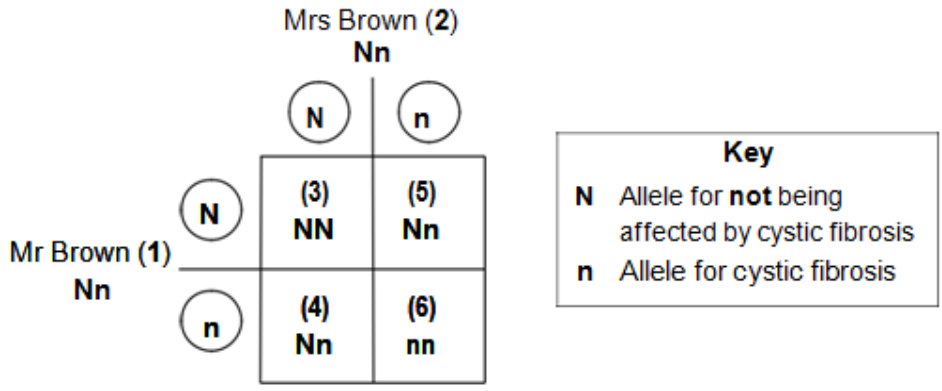
carriers.

immune.

infected.

(1 mark)

(b) The diagram shows how the allele for cystic fibrosis can be inherited by Mr and Mrs Brown's children.



(b) (i) Give the number of one person in the diagram who has cystic fibrosis.

☐

(1 mark)

(b) (ii) The chance that Mr and Mrs Brown's next child will have cystic fibrosis is

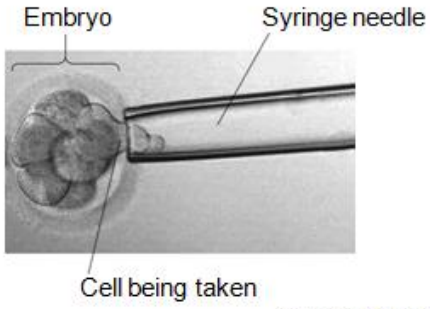
_____

(1 mark)

(c) A genetic counsellor describes to Mr and Mrs Brown one way of screening embryos for cystic fibrosis.

- Some eggs are collected from Mrs Brown.
- The eggs are then fertilised in a dish.
- Several embryos may start to develop.

The photograph shows how doctors take one cell from each embryo when it is only 3 days old.



Source: © Pascal Goetgheluck/Science Photo Library

- The DNA in the cell from each embryo is tested for cystic fibrosis.
- Doctors select one embryo that is unaffected and place it in Mrs Brown's uterus.
- The embryo then develops into a baby.

Use the information to suggest one advantage and one disadvantage of screening embryos in this way.

Advantage _____

_____

Disadvantage _____

_____

(2 marks)

Q:4 A certain gene codes for the production of an enzyme called 'HEXA'.

One human genetic disorder causes damage to nerve cells in the brain. This disorder is caused by a small change in the DNA of the HEXA gene. People with this disorder make a changed HEXA enzyme that does not work.

(a) Explain how a change in the DNA of the HEXA gene can result in the production of a changed HEXA enzyme that does not work.

_____

_____

_____

_____

_____

_____

(3 marks)

(b) The gene coding for the HEXA enzyme is found on chromosome number 15.

(b) (i) How many chromosomes are there in the nucleus of a human nerve cell? _____

(1 mark)

(b) (ii) A boy had the changed HEXA gene on the chromosome number 15 that he inherited from his father.

The changed HEXA gene coded for a HEXA enzyme that does not work. The boy did not develop the genetic disorder.

Explain why the boy did not develop the genetic disorder.

_____

_____

_____

_____

(2 marks)

(b) (iii) The boy grew up and got married.

A blood test showed that his wife had also inherited the same changed HEXA gene.

There is a 1 in 4 chance that this couple's first child will have the genetic disorder.

Use a genetic diagram to explain why.

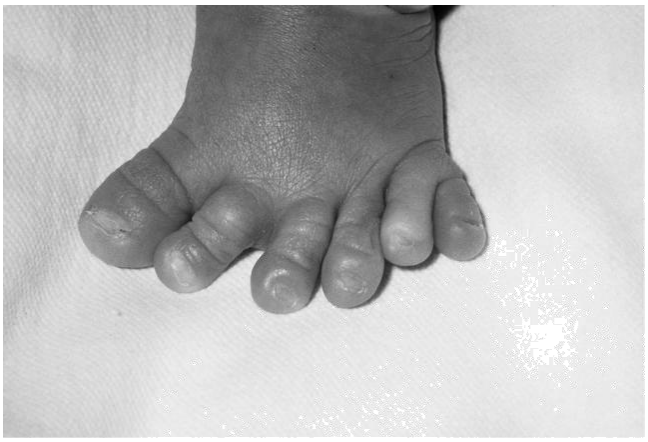
Use the following symbols in your explanation:

H = allele for making the normal HEXA enzyme

h = allele for making a HEXA enzyme that does not work.

(3 marks)

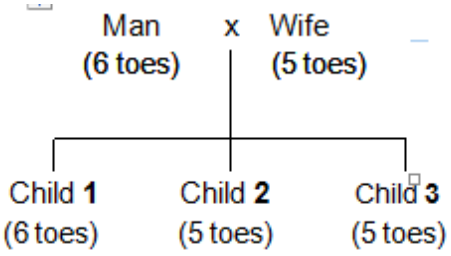
Q:5 Polydactyly is an inherited condition. Polydactyly is controlled by a dominant allele. The photograph shows the foot of a baby with polydactyly.



A man and his wife have three children. The man has polydactyly.

The diagram shows the inheritance of polydactyly in this family.

The diagram also shows the number of toes each person has on each foot.



In the rest of this question, the following symbols are used to represent alleles.

D = allele for polydactyly (6 toes on each foot) d = allele for 5 toes on each foot

(a) (i) How many alleles for the number of toes will there be in one sperm cell?

☐

(1 mark)

(a) (ii) Complete the sentence.

A sperm cell joins with an egg cell in a process called _____

(1 mark)

(b) (i) What combination of alleles does the man have?

Tick (☒) one box.

DD ☐

Dd ☐

dd ☐

(1 mark)

(b) (ii) What combination of alleles does the man's wife have?

Tick (☒) one box.

DD ☐

Dd ☐

dd ☐

(1 mark)

(c) Draw a ring around the correct answer to complete each sentence.

(c) (i) The man and his wife plan to have a fourth child.

The probability that this child will have 6 toes on each foot is

1 in 2.

1 in 3.

1 in 4.

(1 mark)

(c) (ii) When Child 2 grows up, he marries a woman with 5 toes on each foot.

The probability that their first child will have 6 toes on each foot is

0.
1 in 2.
1 in 4.

(1 mark)

Q:7 Phenylketonuria (PKU) is an inherited condition. PKU makes people ill.

(a) PKU is caused by a recessive allele.

(a) (i) What is an allele?

_____

_____

[1 mark]

(a) (ii) What is meant by recessive?

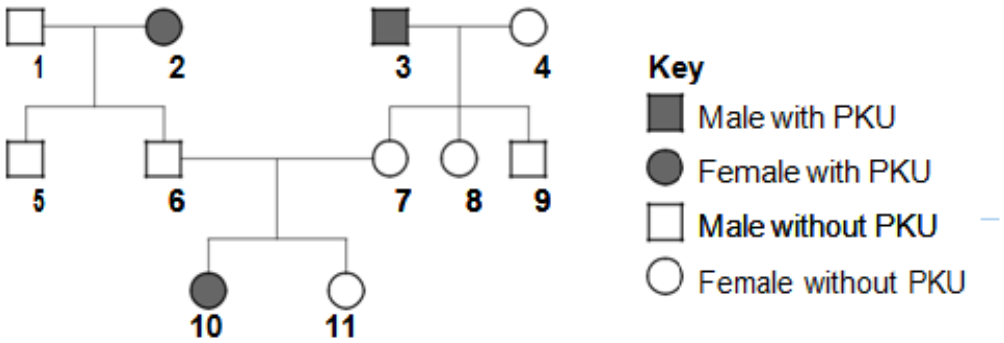
_____

_____

[1 mark]

(b) Figure 8 shows the inheritance of PKU in one family.

Figure 8



(b) (i) Give one piece of evidence from Figure 8 that PKU is caused by a recessive allele.

_____

_____

[1 mark]

(b) (ii) Persons 6 and 7 are planning to have another child.

Use a genetic diagram to find the probability that the new child will have PKU.

Use the following symbols in your answer:

N = the dominant allele for not having PKU n = the recessive allele for PKU.

Probability = _____

[4 marks]

(c) Persons 6 and 7 wish to avoid having another child with PKU.

A genetic counsellor advises that they could produce several embryos by IVF treatment.

(c) (i) During IVF treatment, each fertilised egg cell forms an embryo by cell division.

Name this type of cell division.

_____

[1 mark]

(c) (ii) An embryo screening technique could be used to find the genotype of each embryo.

An unaffected embryo could then be placed in person 7's uterus.

The screening technique is carried out on a cell from an embryo after just three cell divisions of the fertilised egg.

How many cells will there be in an embryo after the fertilised egg has divided three times?

[1 mark]

(c) (iii) During embryo screening, a technician tests the genetic material of the embryo to find out which alleles are present.

The genetic material is made up of large molecules of a chemical substance.

Name this chemical substance.

_____

[1 mark]

(d) Some people have ethical objections to embryo screening.

(d) (i) Give one ethical objection to embryo screening.

_____

[1 mark]

(d) (ii) Give one reason in favour of embryo screening.

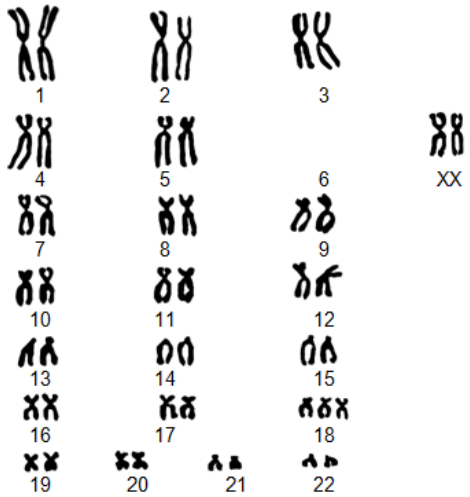
_____

[1 mark]

Q:8 Genetic disorder E is a condition caused by a change in the chromosomes.

(a) Figure 4 shows the chromosomes from one cell of a person with genetic disorder E.

Figure 4



(a) (i) How do you know this person is female?

Use information from Figure 4.

_____

_____

[1 mark]

a) (ii) Describe how the chromosomes shown in Figure 4 are different from the chromosomes from a person who does not have genetic disorder E.

_____

_____

_____

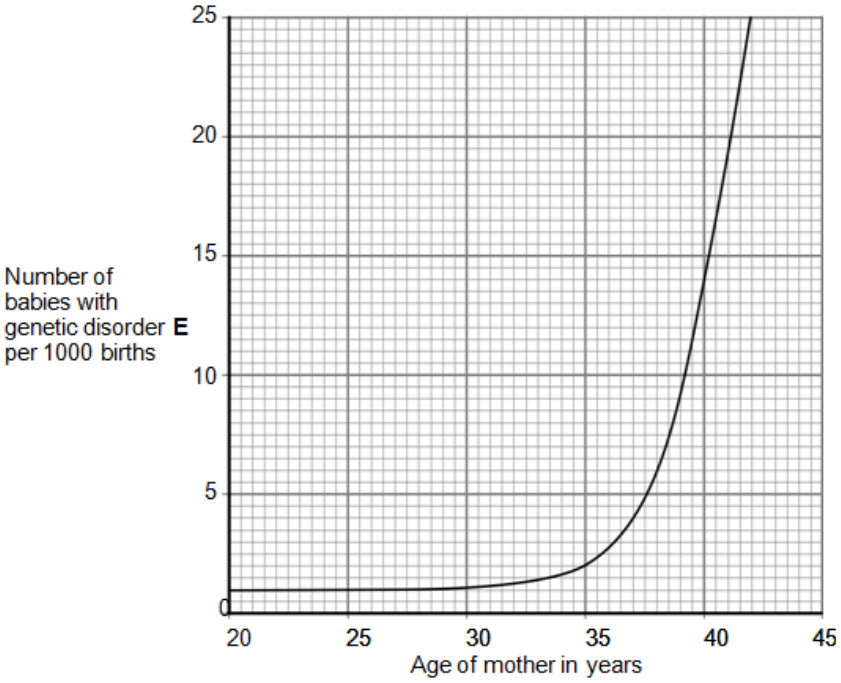
_____

[2 marks]

(b) As a woman gets older, the chance of her having a baby with genetic disorder E increases.

Figure 5 shows this.

Figure 5



(b) (i) The chance of a 35-year-old woman having a baby with genetic disorder E is

2 per 1000 births.

What is the chance of a 40-year-old woman having a baby with genetic disorder E?

..... per 1000 births

[1 mark]

(b) (ii) A 40-year-old woman is more likely than a 35-year-old woman to have a baby with genetic disorder E.

How many times more likely?

_____ times

[1 mark]

(c) A 41-year-old woman wants to have a baby. A 41-year-old woman has an increased chance of having a baby with genetic disorder E.

Doctors can screen embryos for genetic disorder E.

Table 2 gives some information about two methods of embryo screening.

Table 2

Method 1	Method 2
1 The woman is given hormones to cause the release of a few eggs. The eggs are taken from her body in a minor operation. The eggs are fertilised in a glass dish.	1 The woman gets pregnant in the normal way.
2 One cell is taken from each embryo when the embryo is 3 days old.	2 Cells are taken when the embryo is 10 weeks old.
3 Cells are screened for genetic disorder **E**.	3 Cells are screened for genetic disorder **E**.
4 An unaffected embryo is placed in the woman's uterus. Embryos that are not used are destroyed or used in medical research.	4 An unaffected fetus is allowed to develop. If the fetus has genetic disorder **E**, the woman can choose to have an abortion.
5 This method costs about £6000.	5 This method costs about £600.

Use information from Table 2 to give two advantages and one disadvantage of Method 1 compared with Method 2 for detecting genetic disorder E.

Advantages of Method 1:

1 _____

_____

2 _____

_____

Disadvantage of Method 1:

_____

_____

[3 marks]

TOTAL MARKS=60