Questions are for both separate science and combined science students unless indicated in the question

Q1.

Some human disorders are inherited.

Polydactyly is an inherited disorder.

- A person with polydactyly has extra fingers or toes.
- Polydactyly is caused by a dominant allele.

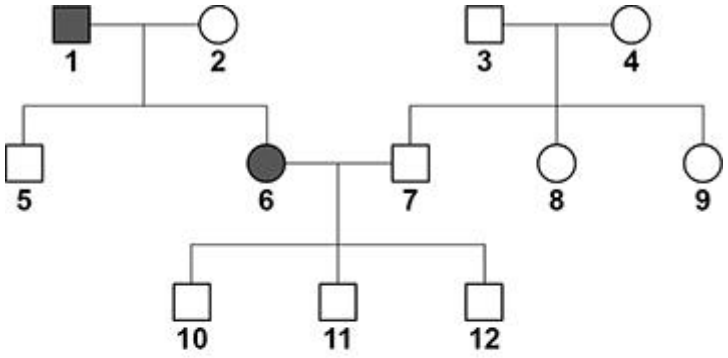
(a) What is a dominant allele?

_____

_____

(1)

The figure below shows the inheritance of polydactyly in one family.



Key

- Male who has polydactyly
- Male who does **not** have polydactyly
- Female who has polydactyly
- Female who does **not** have polydactyly

In part (b) and (c), use the following symbols:

D = allele for having polydactyly

d = allele for **not** having polydactyly.

(b) Person **1** is heterozygous.

Explain how above figure shows that person **1** is heterozygous.

_____

_____

_____

_____

(2)

(c) Persons **6** and **7** are expecting a fourth child.

A doctor states that the probability of having a child with polydactyly is 0.5

Explain how the doctor determined this probability.

You should:

- draw a Punnett square diagram
- give the genotype of person **6** and the genotype of person **7**
- identify **all** the offspring that will have polydactyly.

(HT only)

(4)

(d) Cystic fibrosis (CF) is another inherited disorder caused by a mutation.

The mutation occurs in a gene called CFTR.

For the CFTR gene, one **allele** in every 50 in the UK population is the cystic fibrosis allele.

Explain why only one person in 2500 in the UK population has cystic fibrosis.

(4)

(Total 11 marks)

Q2.

Hormones are important for regulating the menstrual cycle.

During the menstrual cycle, eggs mature inside follicles in the ovaries.

A 27-year-old woman was infertile.

A doctor tested a sample of the woman's blood.

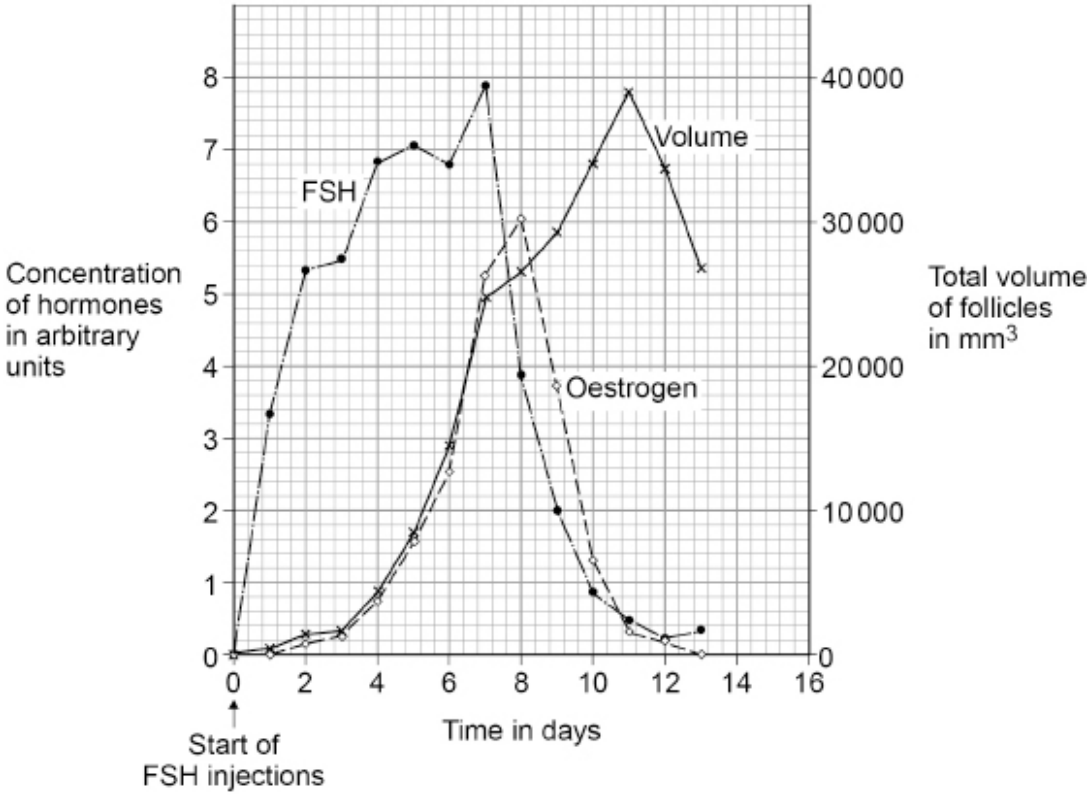
The test did **not** detect any follicle stimulating hormone (FSH) in the woman's blood.

The doctor gave the woman daily injections of FSH for 7 days.

The doctor measured:

- the concentration of FSH in the woman's blood
- the concentration of oestrogen in the woman's blood
- the volumes of developing follicles in the ovaries.

The figure below shows the results.



(a) Give evidence from the figure that the follicles in the ovaries release oestrogen. **(HT only)**

_____

_____

(1)

(b) Injection of FSH caused the development of a number of follicles.

The mean diameter of the follicles on day 11 was 22 millimetres.

Calculate the number of follicles in the woman's ovaries on day 11.

Assume each follicle is a sphere.

Volume of a sphere = $\frac{4}{3}\pi r^3$

r = radius
π = 3.14

Give your answer to the nearest whole number.

Number of follicles (to the nearest whole number) = _____

(5)

(c) Before treatment with FSH, the woman had underdeveloped breasts.

Explain why the lack of FSH in the woman's blood caused underdeveloped breasts.

(2)

(d) Usually males and females both produce FSH.

The woman had inherited a faulty gene for FSH production from each of her parents.

The woman's parents both produce FSH.

Show how the **woman's parents** could have a child that does **not** produce FSH.

You should:
- draw a Punnett square diagram
- identify the phenotype of each offspring genotype
- use the symbols below:
  H = allele for making FSH
  h = allele for **not** making FSH
  (HT only)

(3)

(e) The woman continues to have injections of FSH.

The woman has a child with a man who is heterozygous for the FSH gene.

Explain why the probability that the child will be able to produce FSH is 0.5.

(HT only)

_____

_____

_____

_____

_____

_____

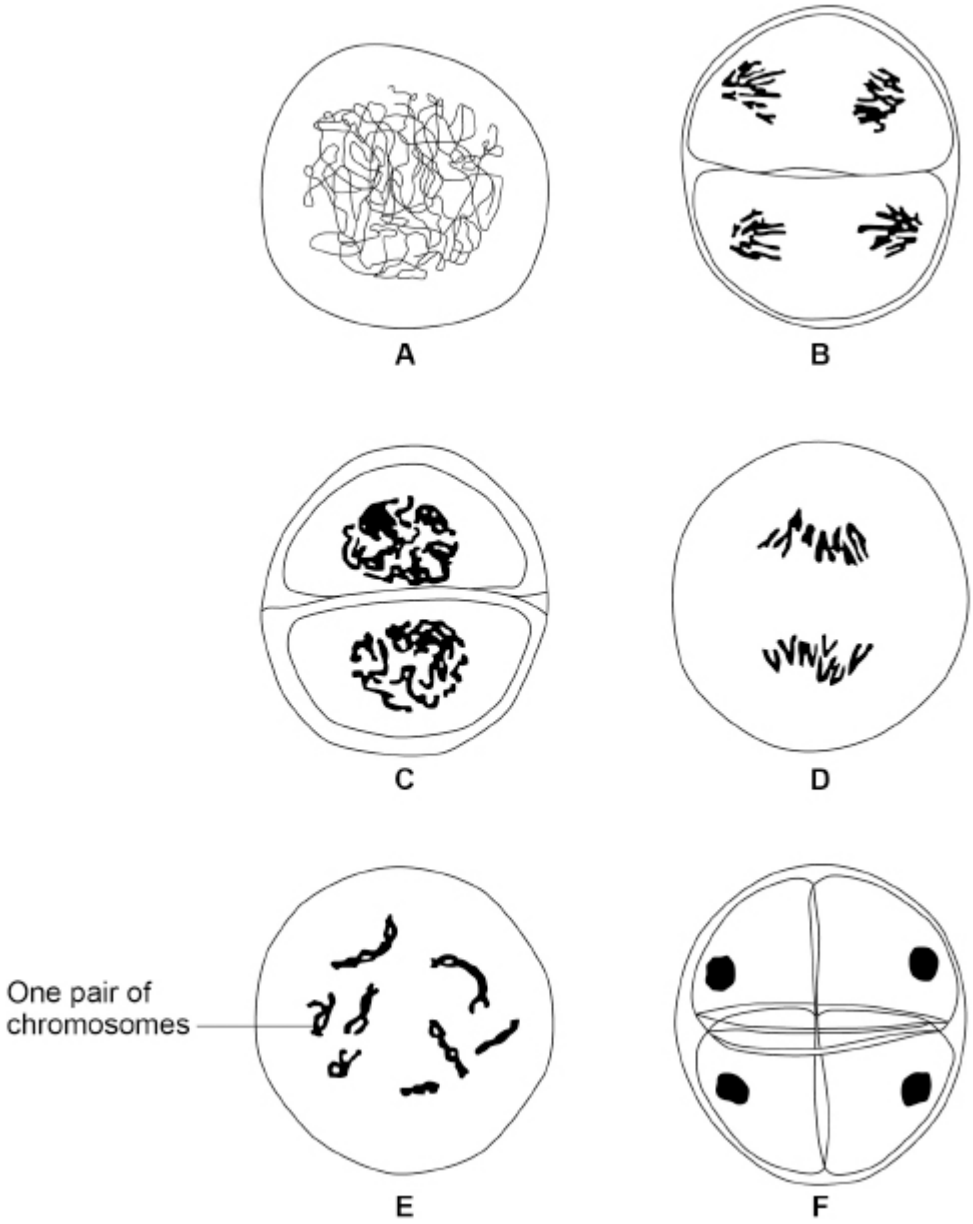
(3)

(Total 14 marks)

Q3.

Figure 1 shows six stages in the process of meiosis.

Figure 1



(a) In **Figure 1**, **A** is the first stage and **F** is the final stage.

Stages **B** to **E** are **not** in the correct order.

Give the correct order of stages **A** to **F**.

A → _____ → _____ → _____ → _____ → **F**

(1)

At the end of meiosis the number of chromosomes is different from the number of chromosomes at the start of meiosis.

(b) Give the number of chromosomes in **one** cell in **Figure 1**:

- at the start of meiosis
- at the end of meiosis.

Start _____

End _____

(2)

(c) Explain why the change in the number of chromosomes is important.

_____

_____

_____

_____

_____

_____

(3)

(d) Meiosis produces cells that are genetically different.

Describe how meiosis produces cells that are genetically different.

_____

_____

_____

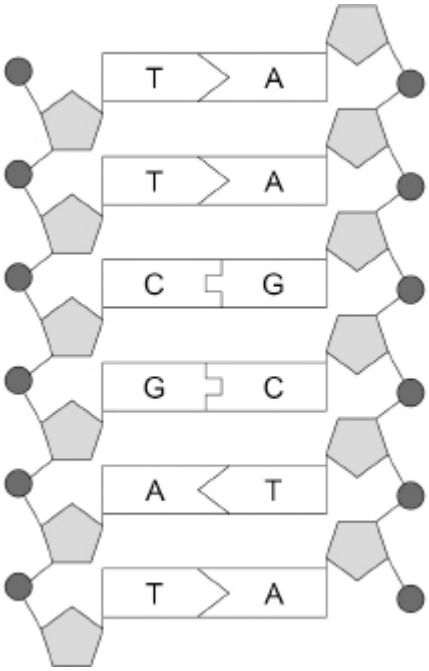
_____

(2)

Chromosomes contain DNA.

Figure 2 shows part of a DNA molecule.

Figure 2



(e) What type of substances are labelled **A**, **C**, **G** and **T** in **Figure 2**? **(biology only)**

_____

(1)

(f) DNA is made of nucleotides.

How many nucleotides are shown in **Figure 2**? **(biology only)**

_____

(1)

(Total 10 marks)

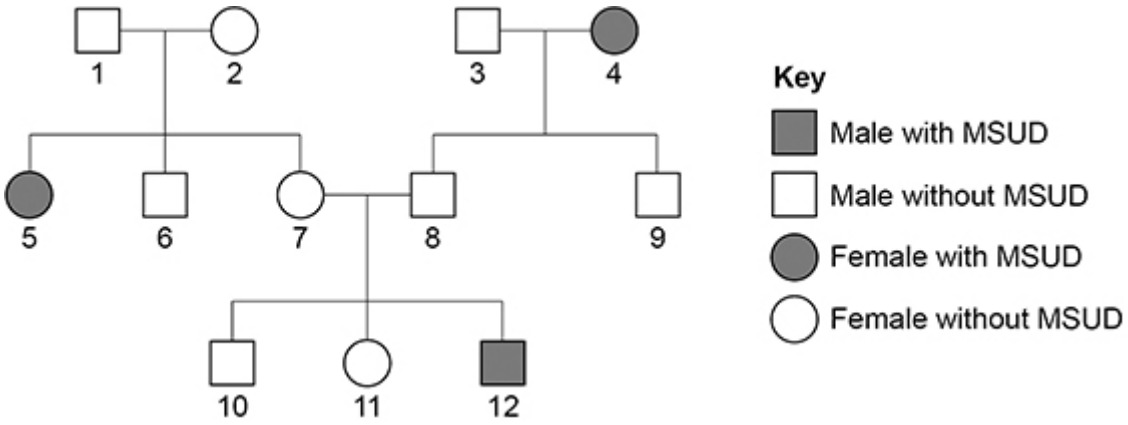
Q4.

Maple syrup urine disease (MSUD) is a rare inherited human condition.

MSUD is usually diagnosed early in childhood and can be controlled by having a low-protein diet.

Figure 1 shows the inheritance of MSUD in one family.

Figure 1



The allele for MSUD is recessive.

(a) Give **one** piece of evidence from **Figure 1** which shows that MSUD is a recessive condition.

_____

_____

(1)

(b) Persons **7** and **8** in **Figure 1** are expecting a fourth child.

Determine the probability that the child will have MSUD.

You should:

- draw a Punnett square diagram
- identify the phenotype of each offspring genotype
- use the symbols:

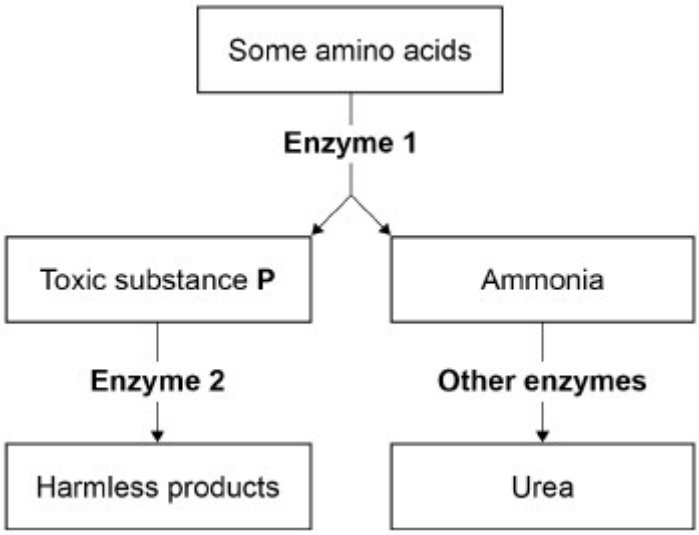
N = allele for **not** having MSUD
n = allele for MSUD.
(HT only)

Probability = _____

(4)

Figure 2 shows chemical reactions involved in the normal breakdown of some types of amino acid inside body cells.

Figure 2



A person with MSUD **cannot** make **Enzyme 2**.

(c) One of the final products shown in **Figure 2** is urea.

Where in the human body are the reactions shown in **Figure 2** most likely to occur?

(biology only)

Tick (✓) **one** box.

Kidney ☐

Liver ☐

Pancreas ☐

Small intestine ☐

(1)

Scientists can analyse blood samples or urine samples to see if a person has MSUD.

The test identifies high concentrations of toxic substance **P**, shown in **Figure 2**.

(d) Explain why the **blood** of a person with MSUD will have a high concentration of toxic substance **P**.

Use information from **Figure 2**. **(biology only)**

(3)

(e) Explain why the **urine** of a person with MSUD will have a high concentration of toxic substance **P**. **(biology only)**

(2)

(f) Explain why a person with MSUD must have a low-protein diet. **(biology only)**

(3)

(Total 14 marks)