Q1.

In cats, males are XY and females are XX. A gene on the X chromosome controls fur colour in cats. The allele **G** codes for ginger fur and the allele **B** codes for black fur.
These alleles are codominant. Heterozygous females have ginger and black patches of fur and their phenotype is described as tortoiseshell female.

The two alleles, **F** and **f** of a different gene, which is **not** sex-linked, interact with the gene controlling fur colour. The allele **F** is dominant and stops the formation of pigment in the fur, resulting in white fur. The allele **f** is recessive and has no effect on fur colour.

(a) Name the type of interaction between the two genes affecting fur colour.

_____

(1)

(b) What phenotype would a cat with the following genotype have?

X^GX^Bff _____

(1)

(c) Complete the genetic diagram to show all the possible genotypes and the ratio of phenotypes expected in the offspring of this cross.

Phenotypes of parents **X^GX^GFf** x **X^BYff**

Genotypes of offspring _____

Phenotypes of offspring _____

Ratio of offspring phenotypes _____

(3)

(d) In a population, 36% of cats had the **F** allele and had white fur.

Use the Hardy–Weinberg equation to calculate the frequency of the **f** allele in this population.

Show your working.

Answer _____

(2)

(Total 7 marks)

Q2.

(a) Give **two** types of cell that can stimulate an immune response.

1 _____

2 _____

(2)

An autoimmune disease causes the immune system to attack healthy body tissues. Scientists investigated the immune responses of healthy mice and mice with autoimmune disease.

The chemical OXA causes an immune response in mice and can make their skin swell. Mice had olive oil applied to their left ear and OXA in olive oil applied to their right ear.

The immune response was recorded in two ways:

- the cellular response by measuring the mean increase in ear thickness 24 hours after exposure to OXA
- the humoral response by measuring the mean concentration of anti-OXA antibody in blood 14 days after exposure to OXA.

Table 1 shows the results of this investigation. The values in the brackets show ± 2 standard deviations. A value of ± 2 standard deviations from the mean includes over 95% of the data.

Table 1

Type of mice	Sex of mice	Mean increase in ear thickness / cm × 10^{-3}	Mean concentration of anti-OXA antibody / arbitrary units
Healthy	Male	17.9 (±4.1)	16 (±3)
	Female	18.5 (±2.9)	14 (±4)
Autoimmune disease	Male	25.9 (±4.5)	14 (±2)
	Female	16.7 (±3.0)	26 (±7)

(b) Suggest and explain **one** reason why olive oil was applied to the left ear of the mice.

_____

_____

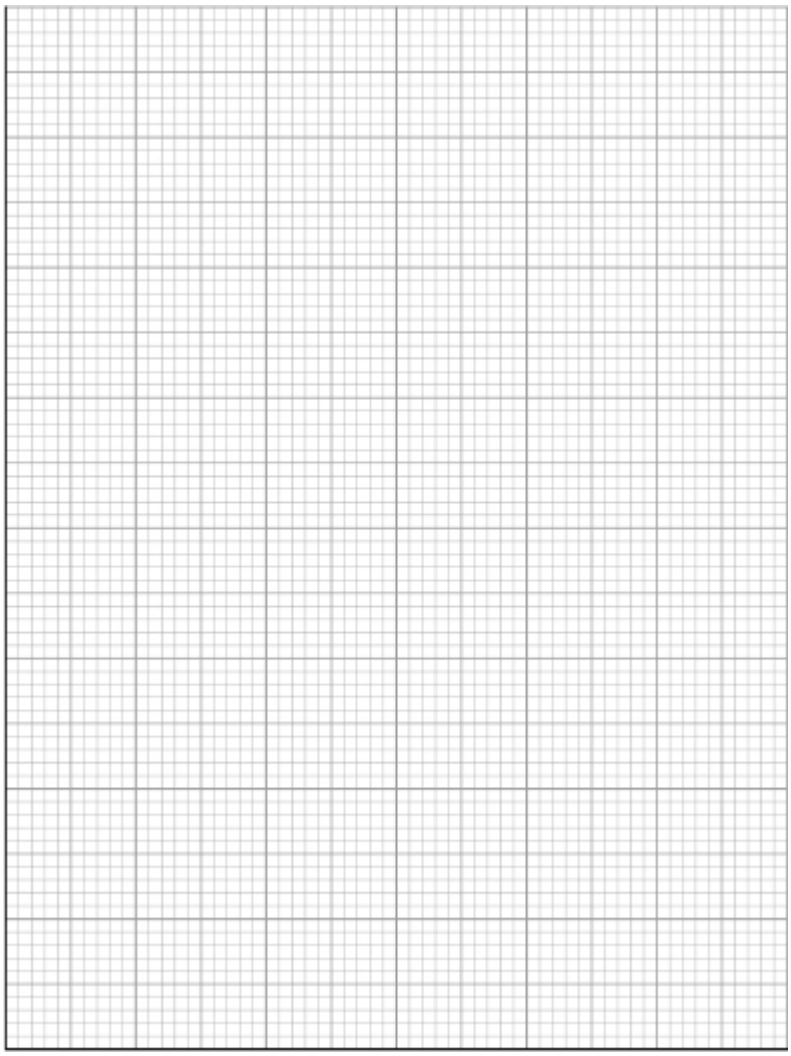
_____

_____

(1)

(c) Plot a suitable graph for mean increase in ear thickness for each group of mice in **Table 1**.

Include the data for ± 2 standard deviations on your graph.



(3)

(d) What can you conclude about the effects of autoimmune disease on the cellular response and the humoral response in male and female mice?

Use the data to justify your conclusions.

(3)

(e) Some studies have shown that **in humans**, oestrogen has the opposite effect on two different autoimmune diseases. Oestrogen:

- accelerates the progression of systemic lupus erythematosus (SLE)
- prevents the progression of rheumatoid arthritis (RA).

The scientists investigated the effect of oestrogen on the immune response in healthy mice and mice with autoimmune disease.

Table 2 shows the scientists' results.

Table 2

Type of mice	Effect of oestrogen on humoral response	Effect of oestrogen on cellular response
Healthy	No effect	No effect
Autoimmune disease	Increase in response	Decrease in response

A student studying these data made the following conclusions.

1. In humans, SLE is caused by an overproduction of antibodies.
2. In humans, RA is caused by an overproduction of cytotoxic T cells (T_C cells).

Evaluate the student's conclusions.

(4)

(f) In mice, one type of autoimmune disease is inherited as a dominant allele. Would the Hardy–Weinberg principle hold true for a population of mice, some of which had this autoimmune disease?

Explain your answer.

(2)

(Total 15 marks)

Q3.

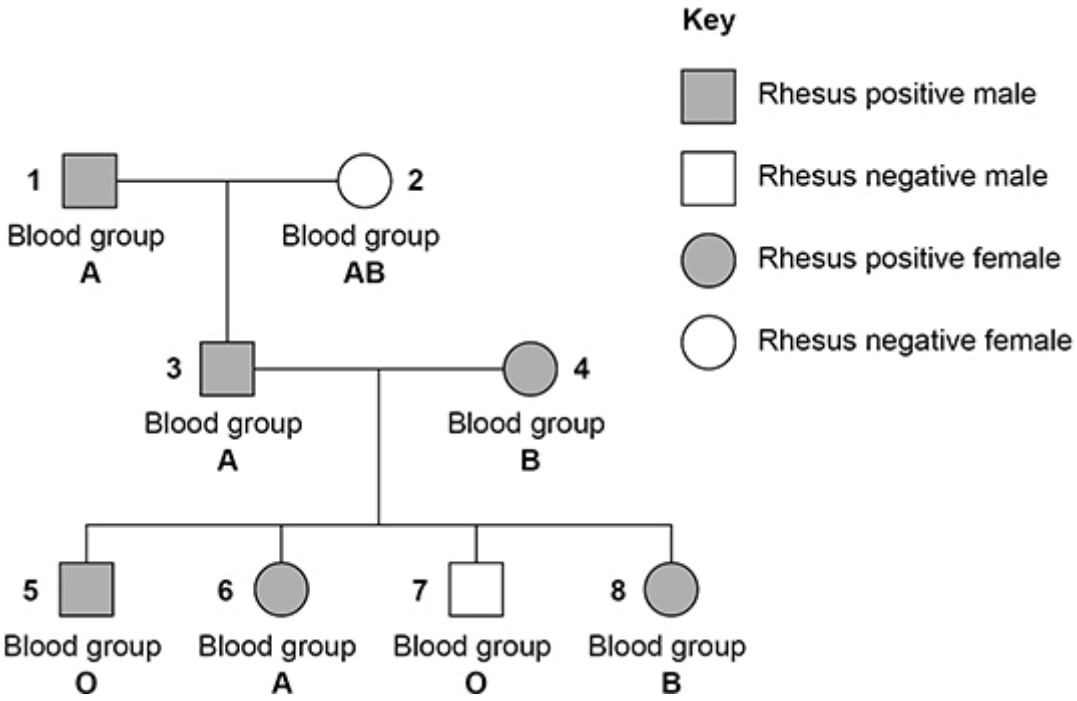
In humans, the ABO blood groups and Rhesus blood groups are under genetic control. The inheritance of the ABO blood groups is controlled by three alleles of a single gene, I^A, I^B and I^O. The alleles I^A and I^B are codominant, and the allele I^O is recessive to I^A and recessive to I^B.

There are four ABO phenotypes, **A**, **B**, **AB** and **O**.

The gene for the Rhesus blood groups has two alleles. The allele for Rhesus positive, **R**, is dominant to the allele for Rhesus negative, **r**.

The genes for the ABO and Rhesus blood groups are **not** sex-linked and are **not** on the same chromosome.

The diagram below shows the phenotypes in a family tree for the ABO and Rhesus blood groups.



(a) Give the genotypes of the ABO blood groups for individuals **1** and **2**.

Do **not** include the genotypes for the Rhesus blood groups in your answer.

1 _____

2 _____

(1)

(b) Explain **one** piece of evidence from the figure above that the allele for Rhesus positive is dominant.

_____

_____

_____

_____

_____

_____

(2)

(c) Calculate the probability of individuals **1** and **2** producing a Rhesus positive son with blood group **A** (individual **3**). You can assume that individual **1** is heterozygous for the Rhesus blood group.

Show your working.

Probability _____

(2)

Scientists determined the frequencies of the ABO alleles and ABO phenotypes in a large population. They then used a statistical test to determine if the frequencies of the four phenotypes differed significantly from the frequencies expected according to the Hardy–Weinberg equation.

(d) The frequencies of the $\mathbf{I^A}$ and $\mathbf{I^O}$ alleles were 0.15 and 0.65. What is the frequency of the $\mathbf{I^B}$ allele?

Frequency of $\mathbf{I^B}$ allele _____

(1)

(e) Name the statistical test you should use to determine if the observed frequencies of the four phenotypes differed significantly from the frequencies expected according to the Hardy–Weinberg equation.

State how many degrees of freedom should apply.

Statistical test _____

Number of degrees of freedom _____

(2)

(f) The scientists concluded that the observed frequencies of the four phenotypes differed significantly from the expected frequencies. Use your knowledge of the Hardy–Weinberg principle to suggest **two** reasons why.

1 _____

_____

_____

2 _____

_____

_____

(2)

(Total 10 marks)