Q1.

Read the following passage.

DNA is a stable molecule but, even under normal cell conditions, spontaneous changes occur to the DNA nucleotide sequence. One example of a spontaneous change occurs when a cytosine base in a guanine–cytosine nucleotide pair is changed to a uracil base. This produces a guanine–uracil nucleotide pair in the DNA molecule. Scientists estimate this type of spontaneous change occurs to 100 guanine–cytosine nucleotide pairs in the genome of healthy human cells every day.

In healthy cells, enzyme-controlled processes repair these spontaneous changes in the DNA molecule by changing uracil bases back to cytosine bases. If these repairs do **not** happen, the uracil DNA nucleotide attracts an adenine DNA nucleotide when the DNA is replicated in the cell cycle. A mutation of the original DNA has now occurred.

Healthy cells with damaged DNA produce enzyme **X**. This enzyme slows the cell cycle by delaying the start of DNA replication. People with the disease ataxia telangiectasia (AT) do not produce functional enzyme **X**. Mutations occur at a higher rate in people with AT.

Use the information in the passage and your own knowledge to answer the following questions.

(a) Give **one** similarity in structure between a guanine–cytosine nucleotide pair and a guanine–uracil nucleotide pair in a DNA molecule (lines 3–5).

Do **not** refer to guanine in your answer.

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(1)

(b) The DNA in a human genome contained 3×10^9 nucleotide pairs.

Assume 40% of these nucleotide pairs are guanine–cytosine nucleotide pairs.

Use this information and lines 5–7 to calculate the percentage of guanine–cytosine nucleotide pairs that change to guanine–uracil nucleotide pairs in this genome every day.

Give your answer in standard form.

Show your working.

Answer _____ %

(2)

(c) The type of mutation that occurs when 'repairs do **not** happen' (lines 10–11) may still produce a functional protein.

Suggest and explain why.

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(4)

(d) Suggest and explain why ‘mutations occur at a higher rate’ in people with AT (lines 15–16).

(3)

(Total 10 marks)

Q2.

(a) Edwards' syndrome is a condition resulting from an extra chromosome 18. A chromosome mutation in the second meiotic division is the most frequent cause of Edwards' syndrome.

Explain how a chromosome mutation in the second meiotic division could result in an extra chromosome 18.

In your answer, **name** the type of chromosome mutation which would result in the extra chromosome.

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(2)

(b) Complete trisomy 18 is the most common type of Edwards' syndrome. This occurs when all the cells of the body have an extra chromosome 18.

Explain why all the cells of the body have an extra chromosome 18.

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(2)

(c) Mosaic trisomy 18 is another type of Edwards' syndrome. This occurs due to a chromosome mutation after fertilisation.

In mosaic trisomy, the body has cells with an extra chromosome 18 **and** cells with the correct number of chromosomes.

Explain how cells with different numbers of chromosomes are produced in mosaic trisomy.

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(1)

(d) The age of the female parent is a factor linked to the risk of a child having Edwards' syndrome.

Which statistical test should be used to test whether this link is statistically significant?

Tick (✓) **one** box.

Correlation coefficient ☐

Chi-squared ☐

Student's t-test ☐

(1)

(e) A ventricular septal defect (VSD) is a common feature of Edwards' syndrome.

A VSD is a hole in the wall between the two ventricles of the heart. A VSD can cause higher blood pressure in the lungs.

Explain how a VSD can cause higher blood pressure in the lungs.

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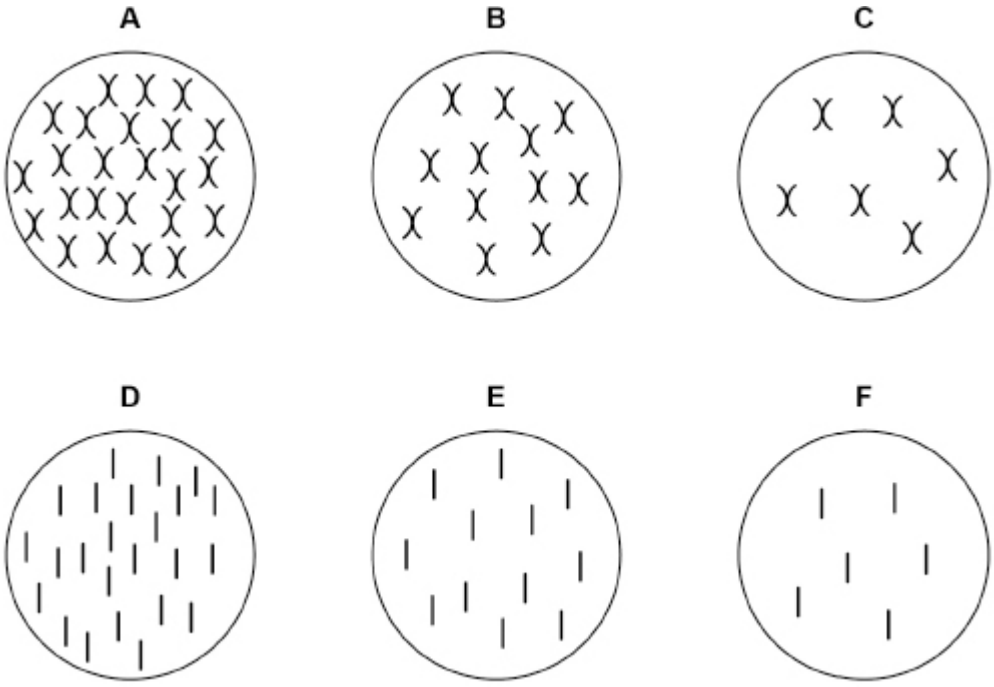
(2)

(Total 8 marks)

Q3.

(a) The figure below shows diagrams of six possible arrangements of chromosomes in cells.



Before meiosis, a cell of a rice plant has 12 pairs of homologous chromosomes (24 chromosomes in total).

Give the letter of the diagram from the figure above that correctly shows the chromosome content of rice cells after the first meiotic division and after the second meiotic division.

After first meiotic division _____

After second meiotic division _____

(2)

(b) Scientists have produced a mutated rice variety in which there is **no** crossing over.

A population of the mutant rice variety produced by sexual reproduction shows genetic variation. Populations of non-mutant rice varieties also show genetic variation.

Suggest and explain the similarities and differences in the causes of genetic variation within these rice populations.

(3)

(Total 5 marks)

Q4.

(a) Describe how a quaternary protein is formed from its monomers.

Do not include the process of translation in your answer.

(5)

(b) Describe the structure of DNA and the structure of a chromosome.

(6)

(c) Mutation can result in an increase in genetic variation within a species.

Describe and explain the **other** processes that result in increases in genetic variation within a species.

(4)

(Total 15 marks)

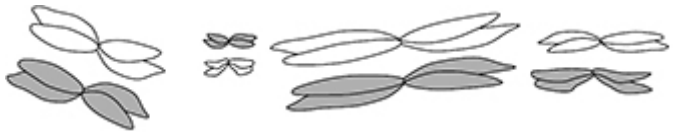
Q5.

(a) Describe how the process of meiosis results in haploid cells.
Do **not** include descriptions of how genetic variation is produced in meiosis.

(4)

(b) **Figure 1** shows the arrangement of chromosomes in a cell during the first meiotic division.

Figure 1



A scientist observed 300 cells. All of the cells were at exactly the same stage of meiosis as the cell shown in **Figure 1**.

Use your knowledge of the independent segregation of homologous chromosomes to calculate how many of these cells are expected to have an **identical arrangement** of chromosomes to those shown in **Figure 1**. Assume no crossing over occurs.

Answer _____

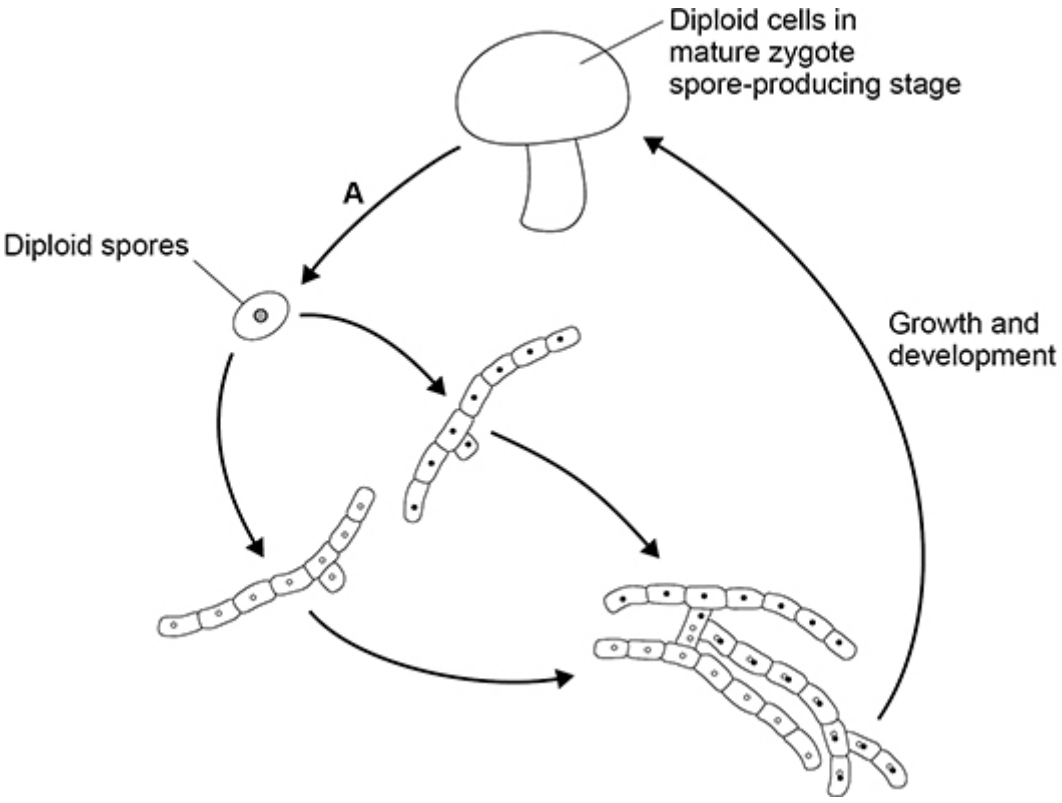
(2)

(c) Draw a diagram to show the chromosomes in one gamete produced by meiosis from the cell shown in **Figure 1**.

(1)

(d) **Figure 2** shows the life cycle of a fungus. The life cycle includes sexual reproduction.

Figure 2



What is the name of the process shown by arrow **A** in **Figure 2**?

Tick (✓) **one** box.

Binary fission ☐

Fertilisation ☐

Meiosis ☐

Mitosis ☐

(1)

(Total 8 marks)

Q6.

(a) Describe viral replication.

(3)

(b) Complete the table below by putting a tick (✓) where the feature is part of a cell cycle involving mitosis or a cell cycle involving binary fission.

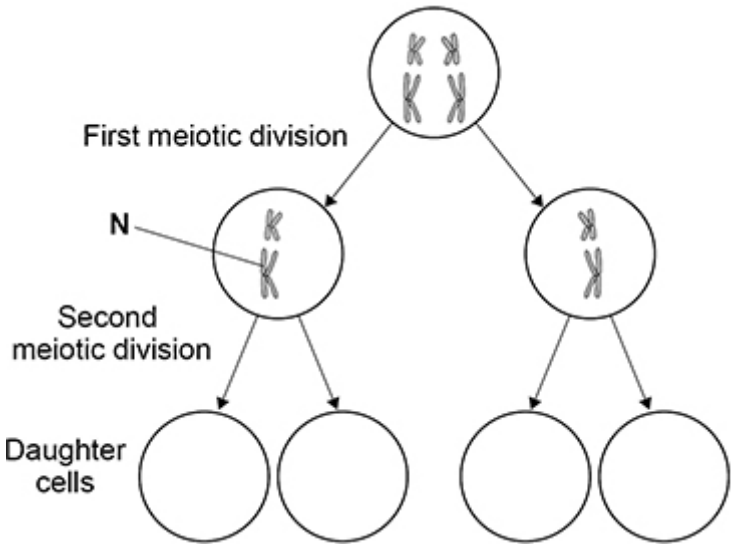
Feature	Cell cycle involving:	
	mitosis	**binary fission**
Replication of linear DNA		
Replication of circular DNA		
Produces 2 daughter cells		
Produces 4 daughter cells		
Happens in prokaryotic cells		
Happens in eukaryotic cells		

(2)

Figure 1 represents a cell undergoing meiosis. It shows the chromosomes in the parent cell and in the two cells formed after the first meiotic division.

The second division of meiosis proceeds normally except that non-disjunction occurs in the chromosome labelled **N**.

Figure 1



(c) Complete **Figure 1** to show the chromosomes inside the daughter cells formed after the second meiotic division.

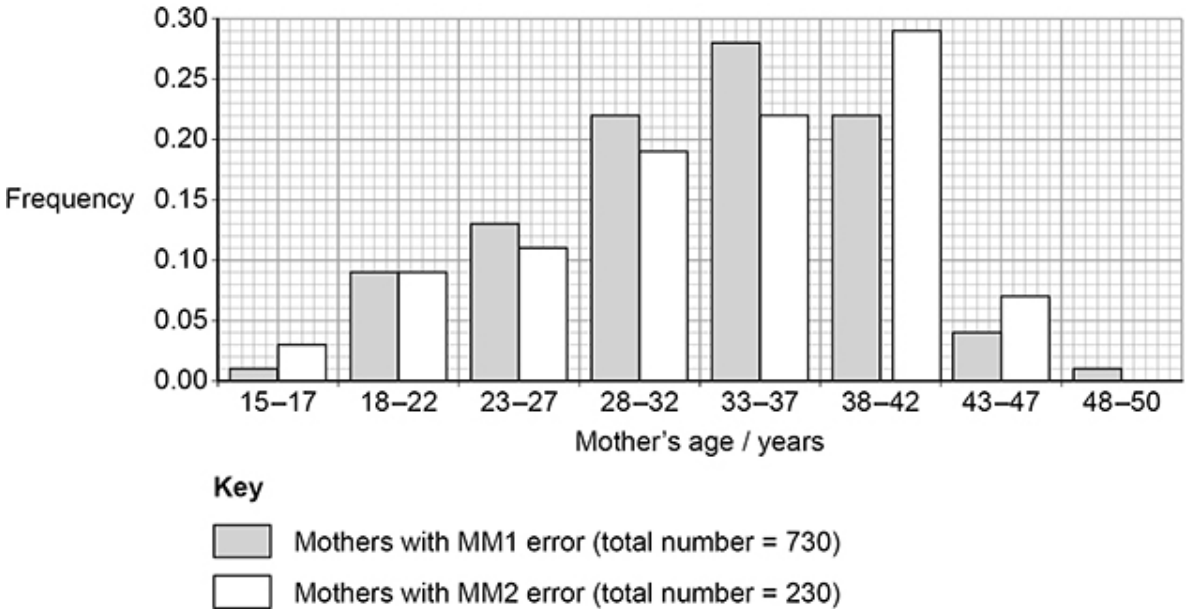
(2)

Doctors studied babies born with a mutation caused by chromosome non-disjunction during gamete formation in their mother.

They determined each mother's age at the time of childbirth and whether the non-disjunction happened in the first meiotic division (MM1 error) or in the second meiotic division (MM2 error).

Figure 2 shows the doctors' results.

Figure 2



(d) A student concluded that there were more mothers of age >37 with MM2 errors than with MM1 errors.

Using **Figure 2** and suitable calculations show why this conclusion is **not** valid.

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(2)

(Total 9 marks)