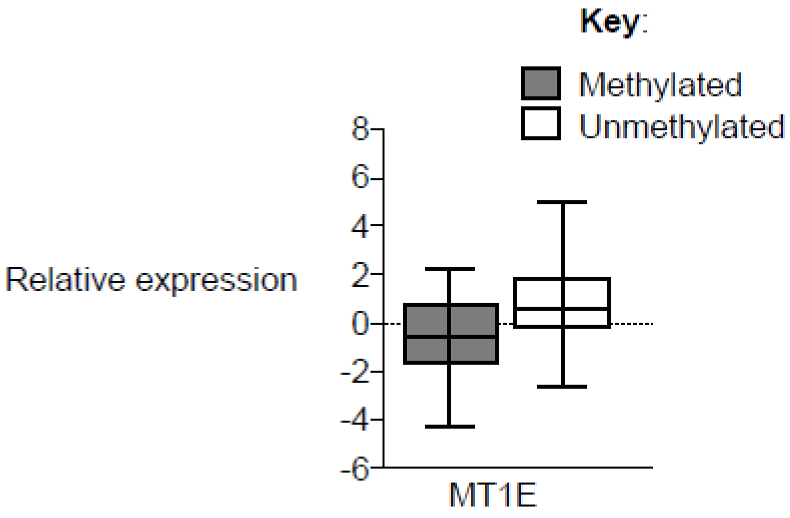
D2.2 Gene expression [64 marks]

1. [Maximum mark: 1]

The graph shows the effect of methylation on the expression of MT1E, a gene involved in the control of prostate cancer development. Patients with a reduced expression of this gene are more likely to develop prostate cancer.



[Source: © 2017 Demidenko et al. 2017. Decreased expression of MT1E is a potential biomarker of prostate cancer progression. *Oncotarget*, 8, pp. 61709–61718. Distributed under the terms of the Creative Commons Attribution License 3.0 (CC BY 3.0). Image redrawn and reannotated.]

What are effects of MT1E methylation?

A. It reduces transcription of MT1E, increasing the risk of prostate cancer.

B. It increases translation of MT1E, reducing the risk of prostate cancer.

C. It reduces replication of MT1E, reducing the risk of prostate cancer.

D. It increases the chances of mutation in proto-oncogenes, increasing the risk of prostate cancer. [1]

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2. [Maximum mark: 1]

Scientists measured methylation levels of amino acids in chromosome 2 in twin studies. The methylation levels of twin 1 were significantly and consistently higher than those of twin 2. What could be proposed to explain this data?

A. Modification to the histone of twin 1

B. Removal of the histone from the chromosome in twin 2

C. The DNA base sequence of twin 1 was altered

D. Cytosines in promoters of twin 2 are more methylated than in twin 1 [1]

3. [Maximum mark: 9]

The table shows the number of amino acids that are different in the sequences of the protein FOXP2 from a human compared with four other primate species.

Primate species	**Number of amino acid differences compared to human**
Pan paniscus	2
Pan troglodytes	2
Gorilla gorilla	2
Pongo pygmaeus	3

[Source: Enard, W., Przeworski, M., Fisher, S.E., Lai, C.S., Wiebe, V., Kitano, T., Monaco, A.P. and Pääbo, S., 2002. Molecular evolution of FOXP2, a gene involved in speech and language. *Nature* 418(6900), pp. 869–72. https://doi.org/10.1038/nature01025. Source adapted.]

(a) Identify the total number of different genera named in the table. [1]

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(b) Deduce whether it is possible to conclude that *Pan paniscus* and *Gorilla gorilla* are more closely related to each other than to humans. [2]

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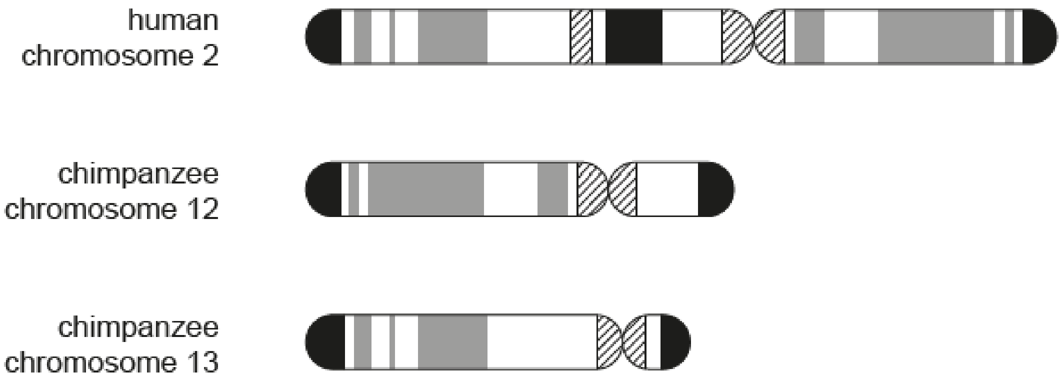
Humans have 46 chromosomes. However, each of the four other primates in the table have 48 chromosomes.

Scientists analysed chromosome 2 in humans and chromosomes 12 and 13 in chimpanzees (*Pan troglodytes*). The diagrams show a summary of their findings.



Key:

■ DNA base sequence usually associated with telomeres

▨ DNA base sequence usually associated with centromeres

(c) Using evidence from the diagrams, justify the hypothesis that chromosome 2 in humans arose from the fusion of chromosomes 12 and 13 with a shared primate ancestor. [4]

(d) Some infectious diseases in humans are caused by viruses that originated in other primates.

(d.i) State **one** structural feature of a virus. [1]

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(d.ii) State the term used for an infectious disease that can transfer from other species to humans. [1]

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4. [Maximum mark: 15]

The genome is the total of all the DNA in an organism, containing all the genetic information required for development and growth.

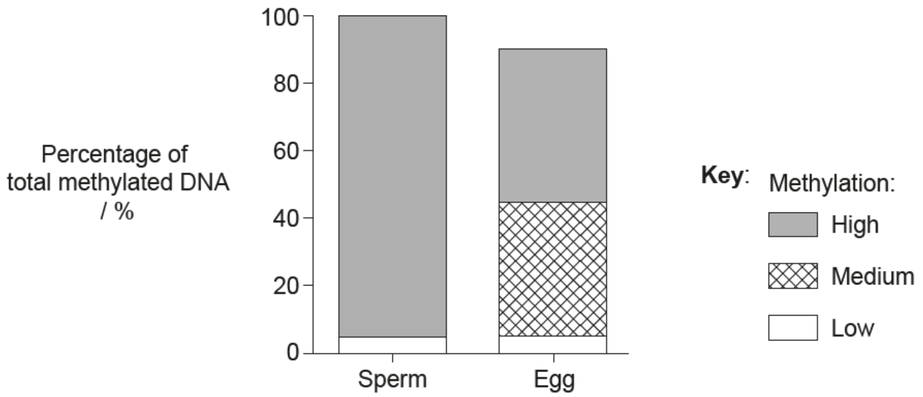
(a) Describe the possible causes and consequences of a substitution mutation in DNA. [4]

(b) Distinguish between cell division by mitosis and by meiosis in eukaryote cells. [4]

(c) Explain how gene expression can be regulated during transcription to determine an organism’s phenotype. [7]

5. [Maximum mark: 1]

DNA methylation profiles in zebrafish (Danio rerio) gametes were determined. The methylated areas were divided into three groups according to the amount of methylation: high, medium and low methylation.



[Source: Potok, M.E., Nix, D.A., Parnell, T.J. and Cairns, B.R., 2013.
Reprogramming the Maternal Zebrafish Genome afterFertilization to
Match the Paternal Methylation Pattern.
Cell, [e-journal] 153(4), pp. 759–772. http://dx.doi.org/10.1016/j.
cell.2013.04.030.]

Methylation of DNA in sperm and egg is removed immediately after fertilization. What is the reason for this?

A. Methylation allows RNA polymerase to join the promoter.

B. It is needed to form homologous pairs of chromosomes.

[1]

C. It allows expression of genes linked to early development.

D. Transcription of promoters only occurs in methylated genes.

6. [Maximum mark: 1]
What is the proteome of an individual?

A. The amino acids unique to an individual making up the proteins in cells

B. The way in which an individual's polypeptides are folded into a three-dimensional structure

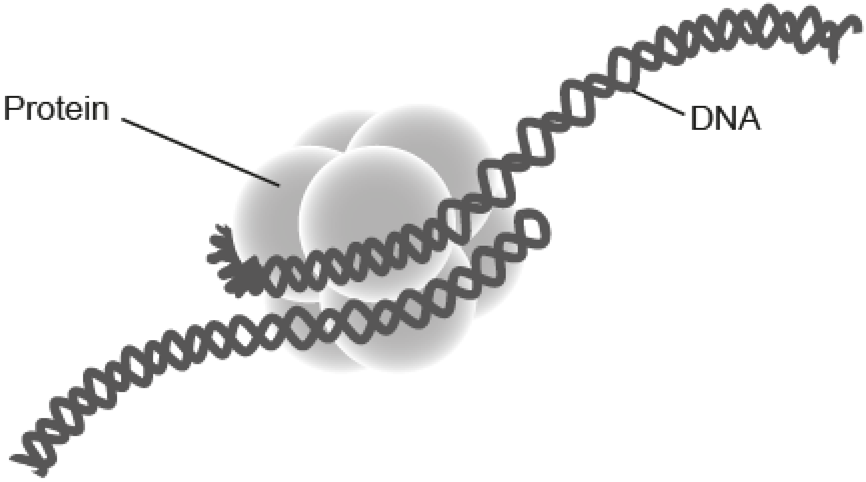
C. The proteins synthesized as an expression of an individual's genes

D. All possible combinations of amino acids an individual contains

[1]

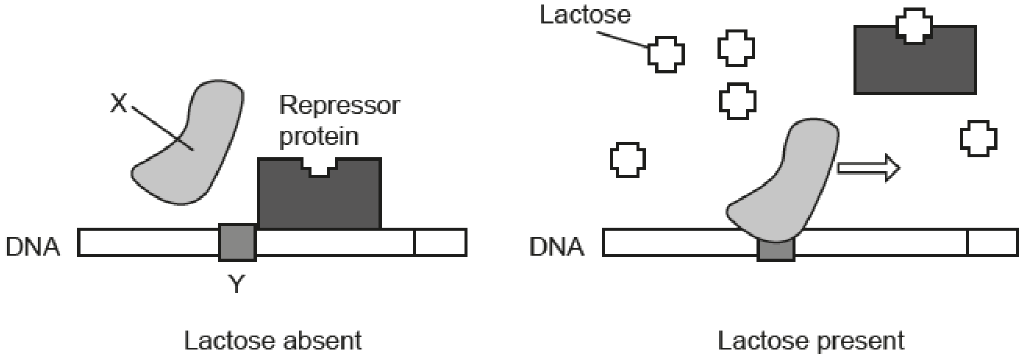
7. [Maximum mark: 8]

The diagram shows a nucleosome from the nucleus of a eukaryotic cell.



[Source: Weissman Lab at UCSF. *UCSF Team Views Genome as it Turns On and Off Inside Cells*.
[diagram online]
Available at https://www.ucsf.edu/news/2011/01/98118/ucsf-team-views-genome-it-turns-and-inside-cells
[Accessed 1 December 2022].]

The image shows the regulation of the gene responsible for producing lactase.



[Source: *Lac Operon*, n.d. [diagram online] T A RAJU. Available at: https://commons.wikimedia.org/wiki/File:Lac_Operon.svg [Accessed 29 October 2021].]

(a) Identify the protein labelled in the diagram. [1]

(b) Outline how nucleosomes affect the transcription of DNA. [1]

(c.i) Identify X, the enzyme which copies a DNA sequence. [1]

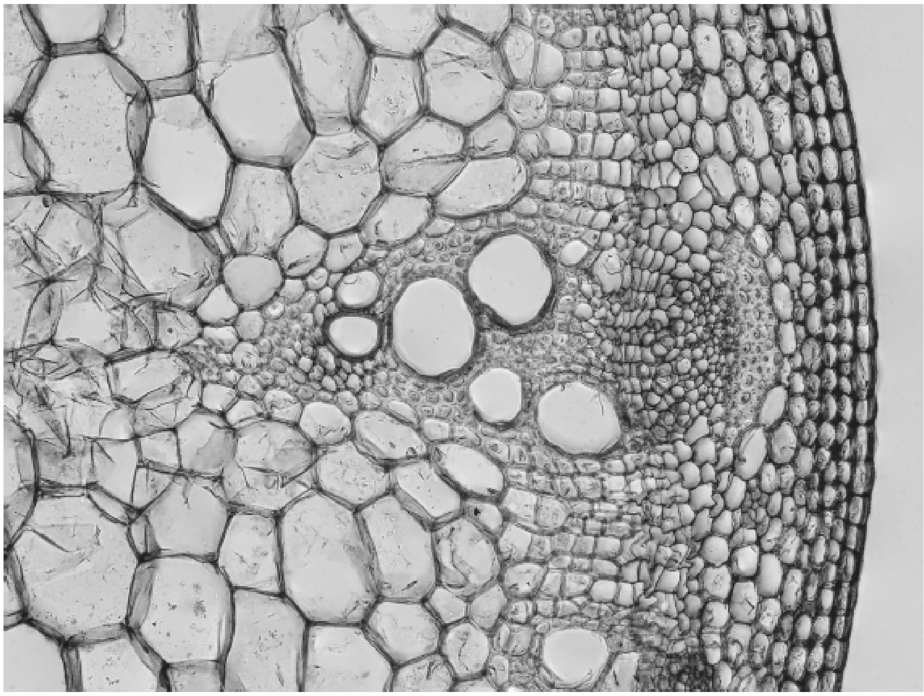
(c.ii) Identify Y, non-coding DNA at the start of a gene. [1]

(d) Explain the role of lactose in the expression of the gene for lactase production. [3]

(e) State **one** reason that identical twins may show different methylation patterns as they grow older. [1]

8. [Maximum mark: 1]

The micrograph of a section through a plant stem shows at least ten different types of cells.



[Source: Joan Carles Juarez / Shutterstock.com.]

What explains the differences between these cells?

A. Only one gene is expressed in each cell type.

B. Different genes are expressed in each cell type.

C. Only useful genes remain in the DNA of each cell type.

D. Changes in the DNA sequence take place when these cells develop. [1]

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9. [Maximum mark: 1]
What is the difference between the DNA of adult identical (monozygotic) twins?

A. Order of genes

B. Sequence of nucleotides

C. Methylation pattern

D. Ratio of complementary base pairs [1]

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10. [Maximum mark: 15]

(a) Describe the structure of the DNA molecule. [5]

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(b) Outline the role of **three** enzymes used in the replication of DNA.

[3]

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(c) Insulin is produced in β cells of the pancreas and not in other cells of the human body. Explain how differentiation of cells and regulation of gene expression allow proteins such as insulin to be produced in only certain types of body cell. [7]

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11. [Maximum mark: 1]

The number of protein-coding genes in the human genome is estimated to be about 20 000, which is much less than the size of the proteome. What is one reason for this?

A. Exons are removed from RNA before translation.

B. There are more types of amino acids than nucleotides.

C. mRNA can be spliced after transcription.

D. Base substitutions occur during transcription. [1]

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12. [Maximum mark: 1]

What is a proteome?

A. The genes that code for all the proteins in the ribosome

B. The group of proteins that generate a proton gradient in mitochondria

C. The entire genome of a prokaryote

D. The entire set of proteins expressed by an organism at a certain time [1]

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13. [Maximum mark: 1]

Which statement correctly describes genome and proteome?

A. Only the genome but not the proteome can be analysed using gel electrophoresis.

B. The genome and the proteome are the same in all tissues in an organism.

C. In cells of different tissues, the genome is the same while the proteome varies.

D. Only mutations in the proteome but not in the genome cause any variability.

[1]

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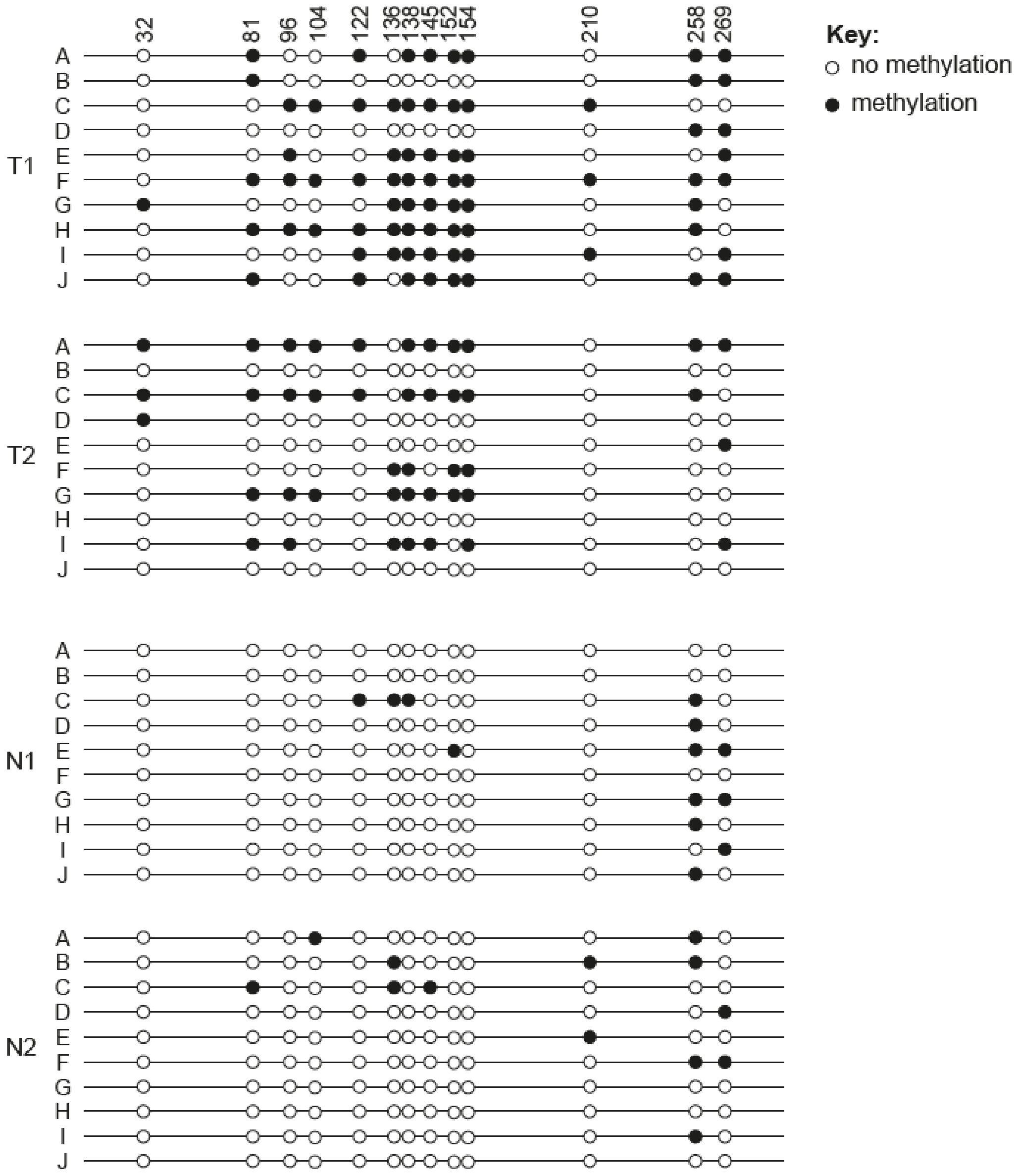
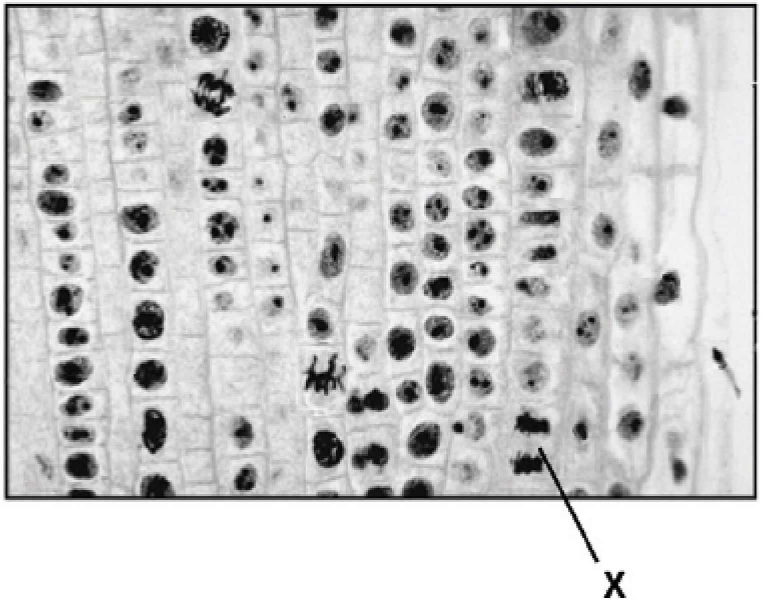
14. [Maximum mark: 8]

DNA methylation has a critical role in gene regulation by affecting transcription. Samples were taken from two colon cancer tumours (T1 and T2) and two normal colon samples (N1 and N2). A particular gene was implicated as a possible cause of cancer. The promoter of this gene was cloned (A–J). The data show the DNA methylation patterns from these samples. The numbers (32–269) represent different markers in the promoter.



[Source: Philipp Schatz, Dimo Dietrich & Matthias Schuster. Rapid analysis of CpG methylation patterns using RNase T1
cleavage and MALDI-TOF. *Nucleic Acids Research* (2004) **32** (21): e167, doi:10.1093/nar/gnh165.
Reproduced by permission of Oxford University Press]

(a.i) Identify the stage of mitosis labelled X in the image, giving a reason.



[1]

[Source: Copyright 2002, The Trustees of Indiana University]

(a.ii) Outline what is indicated by the mitotic index of tissue taken from a tumour. [2]

(a.iii) DNA has regions that do not code for proteins. State **two** functions of these regions.

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2.

[2]

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(b.i) Outline the difference in methylation pattern between tumorous and normal tissue samples.

[2]

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(b.ii) Suggest a way methylation may affect tumour cell genes. [1]

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