



GCE Biology

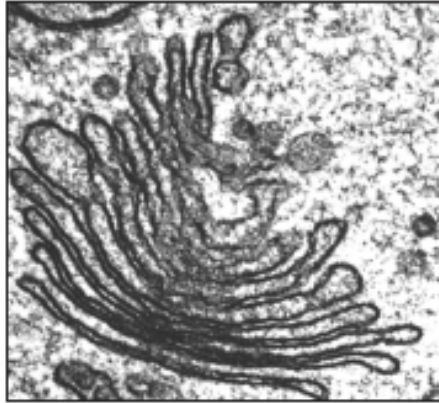
S21-A400U20-1

Assessment Resource 10

Continuity of Life Resource A

Answer all questions.

1. Lysosomal storage disorders are a group of about 50 diseases that are characterised by an accumulation of waste products in the lysosomes. Two examples are Fabry disease and Tay-Sachs disease. Sufferers of Tay-Sachs disease die in childhood.

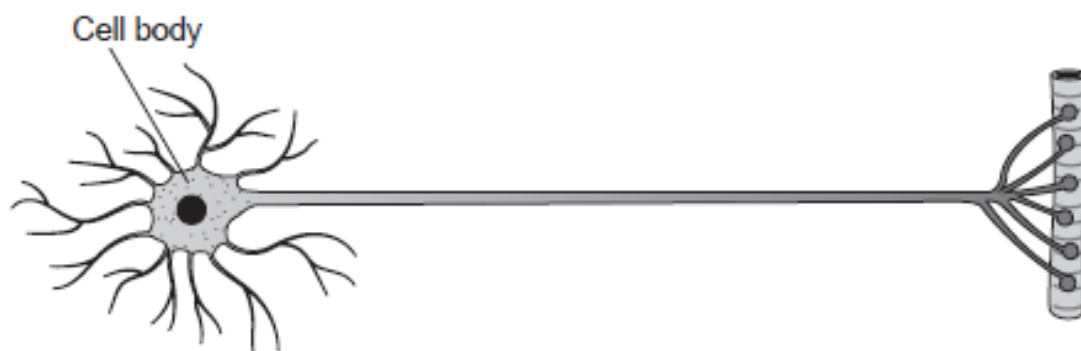


- (a) The electron micrograph above shows the cell organelle responsible for producing lysosomes. Name the organelle, draw an arrow labelled L on the micrograph to identify a lysosome and describe a general function of lysosomes in normal cells. [3]

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- (b) The symptoms of Tay-Sachs disease are a consequence of abnormal accumulation of fatty substances in neurones.

- (i) Complete the diagram of a neurone below to show how this fatty substance is usually distributed, your labels should include the name of the fatty substance. [2]



- (ii) Explain how the arrangement of the fatty substance affects the transmission of nerve impulses. [3]

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- (c) The diagrams below show gene maps of human sex chromosomes and chromosome 15. The recessive allele (**f**) that causes Fabry disease is carried on the X chromosome. The recessive allele (**h**) that causes Tay-Sachs is carried on chromosome 15, in the locus labelled **h**.



- (i) Mark, by placing the letter **f** on the diagram, a possible locus for the gene that causes Fabry disease. [1]

- (ii) With reference to the positions of the gene loci explain the following observations: [2]

- I. Males with the **f** allele always suffer Fabry disease but females can have the **f** allele without suffering the disease.

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- II. Males and females can have the **h** allele without suffering Tay-Sachs disease.

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- (d) Complete the genetic diagram below to show how Fabry disease might be inherited from parents neither of whom suffers from the condition and state the probability of this couple producing a child with Fabry disease. [4]

	male		female
Parental phenotype:
Parental genotypes:	x
Gametes:	x

Probability of this couple producing a child with Fabry disease =

- (e) A genetic counsellor advised a woman with Fabry disease in her family to have an amniotic fluid test, ten weeks into her pregnancy, in order to determine the sex of the embryo.

- (i) Explain the function of amniotic fluid. [1]

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- (ii) Why might it be important for the woman with Fabry disease in her family to know the sex of the embryo and what ethical issues might this cause? [2]

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The Hardy-Weinberg Principle states that the frequency of alleles for a given gene remains constant from generation to generation, providing the population is large and no selection takes place.

The frequency of Tay-Sachs disease is 1 in 360 000 births in the general population of the USA. However, the frequency is 1 in 40 000 in certain populations, which isolate themselves culturally. One such population is the Old Order Amish of the Kishacoquillas Valley, Pennsylvania, USA. Recent estimates put the size of this population at 40 000.

- (f) (i) Use the Hardy Weinberg equations, given below, to calculate the number of people in the Amish community in the Kishacoquillas Valley that carry the Tay-Sachs allele without suffering the disease. [3]

$$p + q = 1.0$$

$$p^2 + 2pq + q^2 = 1.0$$

Number of people who are carriers of Tay-Sachs allele =

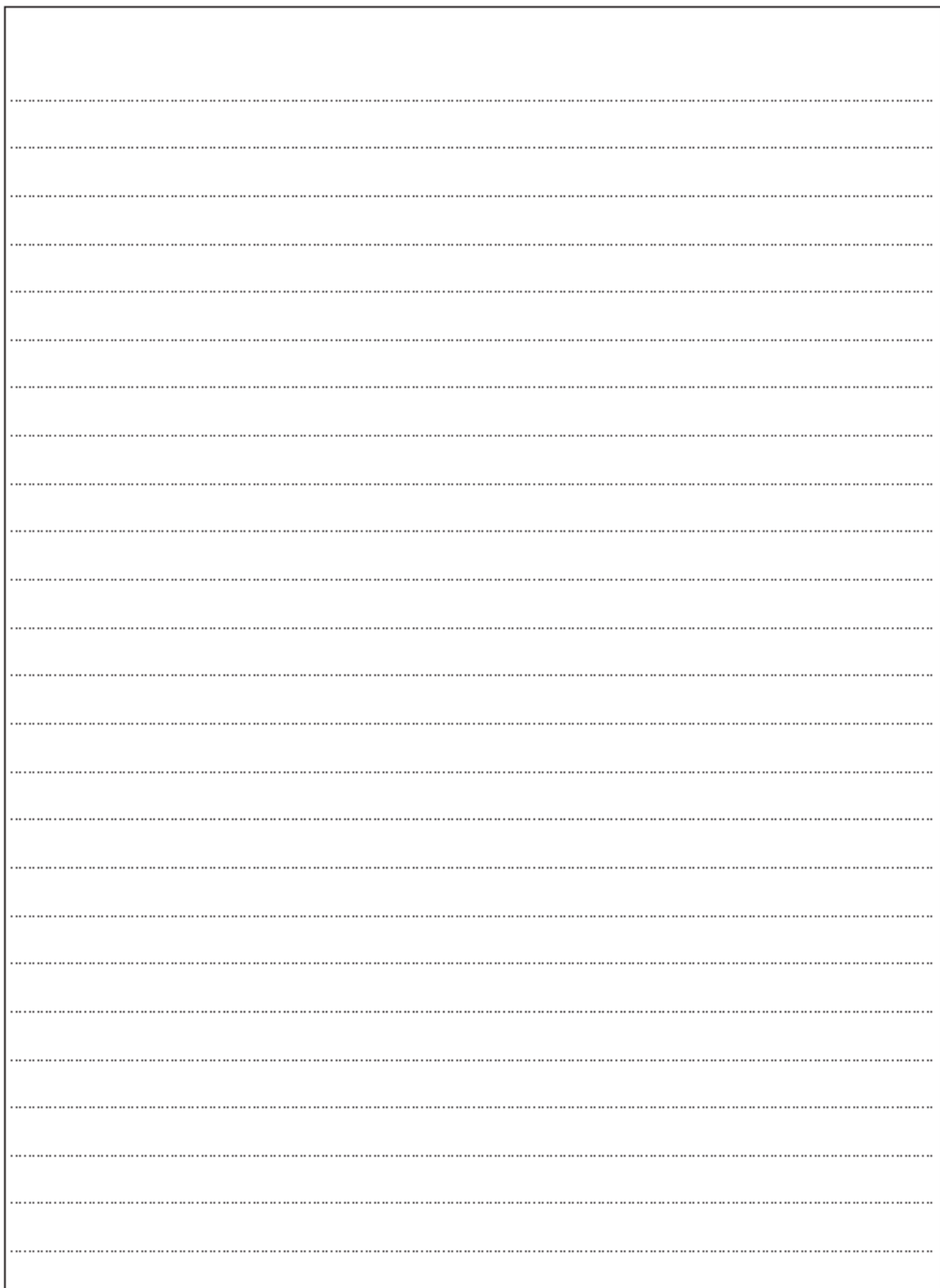
- (ii) Explain why the frequency of Tay-Sachs is higher in isolated populations and predict, with a reason, what is likely to happen to the frequency of the Tay-Sachs allele in the general population. [2]

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Handwriting practice area with horizontal dotted lines.

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