



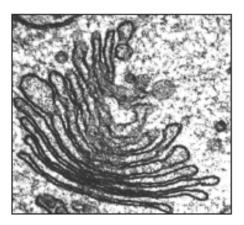
## **GCE Biology**

S21-A400U20-1

## **Assessment Resource 10**

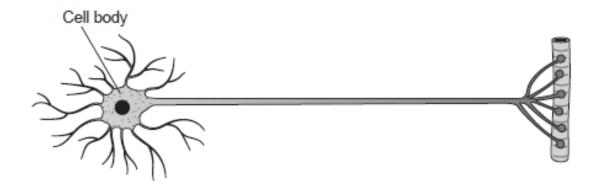
Continuity of Life Resource A

 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]

- (b) The symptoms of Tay-Sachs disease are a consequence of abnormal accumulation of fatty substances in neurones.
  - 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]
(c)	The The rece	diagrams below show gene maps of human sex chromosomes and chromosome 15. recessive allele (f) that causes Fabry disease is carried on the X chromosome. The ssive allele (h) that causes Tay-Sachs is carried on chromosome 15, in the locus lled h.
		X 15
	(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]
		Males with the f allele always suffer Fabry disease but females can have the f allele without suffering the disease.
		II. Males and females can have the h allele without suffering Tay-Sachs disease.

(d)	pare	plete the genetic onto neither of who lucing a child with	m suffers from the con	how Fabry diseas dition and state the	e might be inherited from probability of this couple [4]		
			male		female		
	Pare	ental phenotype:		••••			
	Pare	ental genotypes:		<b>x</b>			
	Gan	netes:		<b>x</b>			
	Prob	pability of this coup	le producing a child wi	th 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 funct	tion of amniotic fluid.		[1]		
	(ii)		important for the woman nbryo and what ethical		ase in her family to know 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 360000 births in the general population of the USA. However, the frequency is 1 in 40000 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 40000.

 (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.

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

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.	In April 2003 one of the most significant scientific breakthroughs of modern times was announced. After years of painstaking research carried out by thousands of dedicated scientists across the world, the complete genetic code of a human being – their genome – could now be made freely available on-line.						
	The Human Genome Project, as this work was known, was the largest international collaboration ever undertaken in biology with British scientists leading the global race to read the human genome using a technique called sequencing.						
	To bring the predicted benefits of genomics to NHS patients the 100 000 Genomes Project was launched in late 2012 and by 2017 had sequenced the genomes of 100 000 NHS patients. The project focussed on patients with a rare disease and their families, and on patients with cancer.						
	Scenario:						
	It is 2025 and Sharon has a painful skin infection that she just can't get rid of. Her doctor would like to prescribe an antibiotic called phenyloxacillin, since it is especially effective against the bacteria (Staphylococcus aureus) that are causing the infection. However, her doctor knows that in a small number of cases phenyloxacillin can cause serious liver damage so suggests a genome test. She tells Sharon that there is a law giving people the right not to disclose the results of genetic tests to insurers.						
	Explain what is meant by 'sequencing' the human genome and describe the type of data that might be made available on-line.  Explain how the extra information provided by the '100 000 Genomes Project' might be used in medicine, and describe how the scenario above illustrates one possible beneficial application and an ethical dilemma of genome sequencing.  [9 QER]						

