

1 Cystic fibrosis and albinism are examples of recessive genetic disorders.  
Tay-Sachs disease is another example of a recessive genetic disorder.

(a) Explain the meaning of the term **recessive genetic disorder**.

(2)

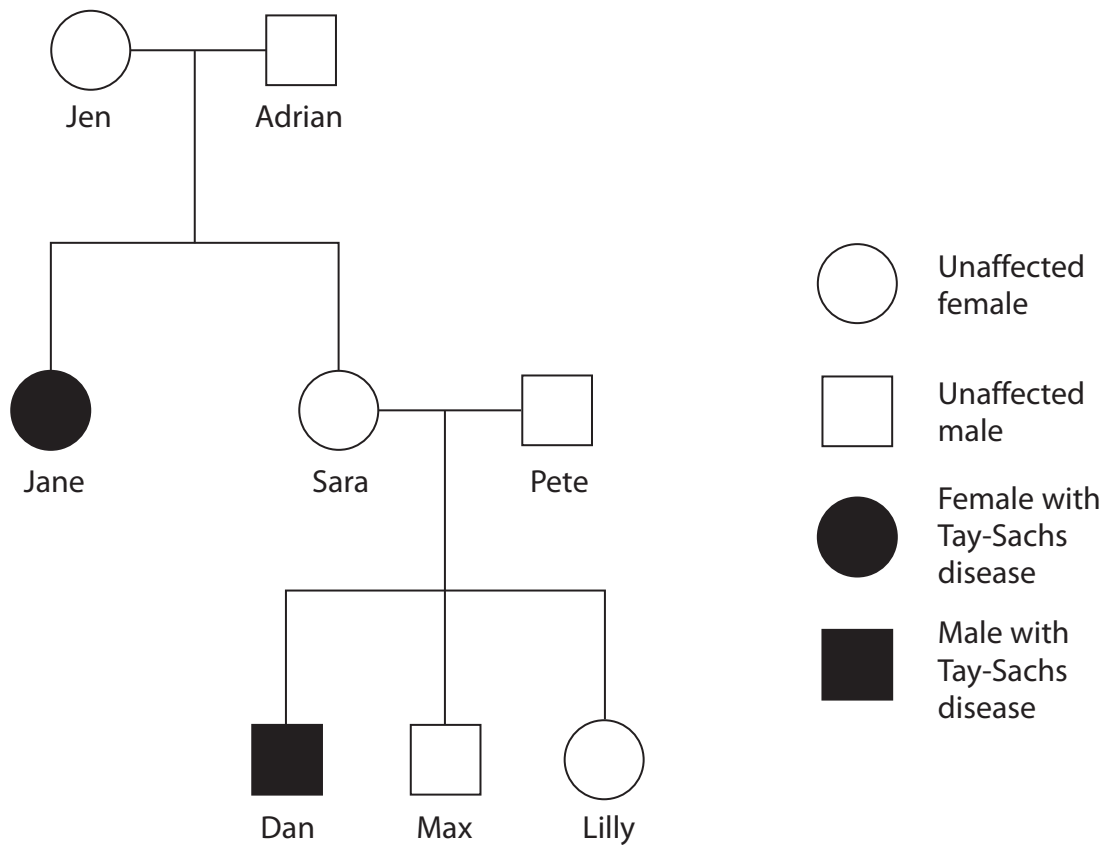
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(b) The genetic pedigree diagram below shows the inheritance of Tay-Sachs disease in one family.



For each of the statements below, put a cross (☒) in the box that correctly completes the statement.

(i) The female who definitely has a homozygous genotype is (1)

**A** Jane

**B** Jen

**C** Lilly

**D** Sara

(ii) The female whose genotype cannot be identified from the diagram is (1)

**A** Jane

**B** Jen

**C** Lilly

**D** Sara

(iii) A male who definitely has a heterozygous genotype is (1)

**A** Adrian

**B** Dan

**C** Max

**D** none of them

(iv) A male who definitely is homozygous dominant is (1)

**A** Adrian

**B** Dan

**C** Max

**D** none of them



**2** Cystic fibrosis and albinism are examples of recessive genetic disorders. Krabbe disease is another example of a recessive genetic disorder. Krabbe disease is caused by mutations in the GALC gene, resulting in a deficiency of an enzyme called galactocerebrosidase.

(a) Explain the meaning of each of the following terms.

(i) Mutation

(2)

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(ii) Recessive

(1)

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(b) Suggest how a mutation in the GALC gene could result in a change in the enzyme galactocerebrosidase.

(3)

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(c) Two parents are both carriers of the recessive allele for Krabbe disease.

In the space below, draw a genetic diagram to show the possible genotypes and phenotypes of their children.

Use the genetic diagram to find the probability of these parents having a child with Krabbe disease.

(5)

Probability .....

(d) State how these parents could determine whether or not their unborn child has Krabbe disease.

(1)

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**(Total for Question 2 = 12 marks)**

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- 3 A study examined the risk of developing a mental disorder. This study determined the risk for both the population as a whole and for those who had a close relative (parent, brother, sister or child) with the same disorder.

The results are shown in the table below.

Mental disorder	Risk of developing mental disorder (%)		
	Population as a whole		Those with a close relative with the same disorder
	Males	Females	
Alcoholism	7.0	2.0	15
Anxiety	3.0	6.0	15
Manic depression	2.0	3.0	15
Neurotic depression	6.0	12.0	11
Obsessive compulsive	0.1	0.1	10
Schizophrenia	1.0	1.0	10

- (a) (i) People with obsessive compulsive disorder (OCD) have symptoms such as repeated washing, checking, touching, counting or arranging.

Using the data in this table, give the evidence that OCD is an inherited condition.

(2)

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(ii) Using the data in the table, explain the validity of the statement that 'OCD is an inherited condition'.

(2)

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(iii) Using the data in the table, state which disorder is least likely to be an inherited condition.  
Give a reason for your answer.

(2)

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**(Total for Question 3 = 6 marks)**





(b) If the phenotypes of the parents are known, the probabilities of having a child with beta thalassaemia, an unaffected child or a child who is a carrier, can be calculated.

Complete the table below to show the results of these calculations.

(4)

<b>Parent 1</b>	<b>Parent 2</b>	<b>Probability of having a child with beta thalassaemia</b>	<b>Probability of having an unaffected child</b>	<b>Probability of having a child who is a carrier</b>
Unaffected	carrier	no chance	50%	50%
Carrier	carrier			
Unaffected	has beta thalassaemia			
Carrier	has beta thalassaemia	50%	no chance	50%

