Questions

Q1.

Hereditary spherocytosis is a condition that affects red blood cells.

Most cases of hereditary spherocytosis are caused by a dominant allele.

Use a genetic diagram to determine the probability of a child inheriting this condition if one parent is heterozygous and the other parent does not have the condition.

(2)

Answer .....

(Total for question = 2 marks)

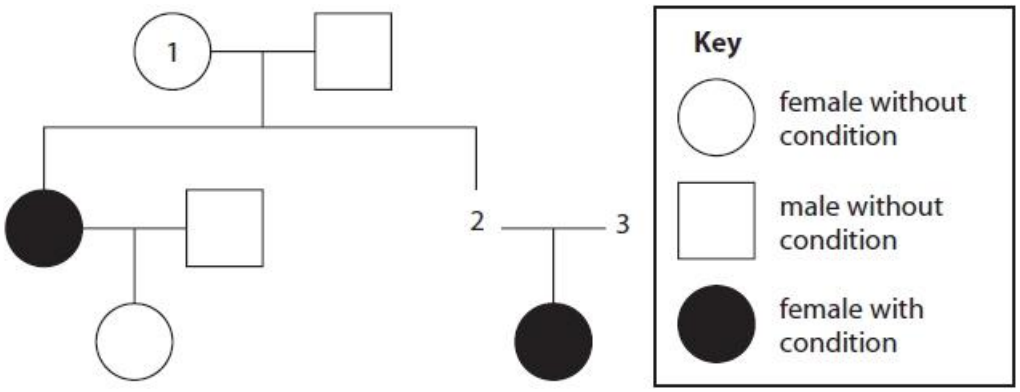
Q2.

Mucopolysaccharides are complex molecules found in the human body.

Mucopolysaccharidosis type 1 (MPS 1) is a recessive genetic condition.

People with MPS 1 cannot break down mucopolysaccharides.

The pedigree diagram shows the inheritance of MPS 1 in a family.



Determine the probability that person 2 has the same sex and MPS 1 phenotype as person 1.

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(Total for question = 4 marks)

Q3.

Answer the question with a cross in the box you think is correct ☒. If you change your mind about an answer, put a line through the box ☒ and then mark your new answer with a cross ☒.

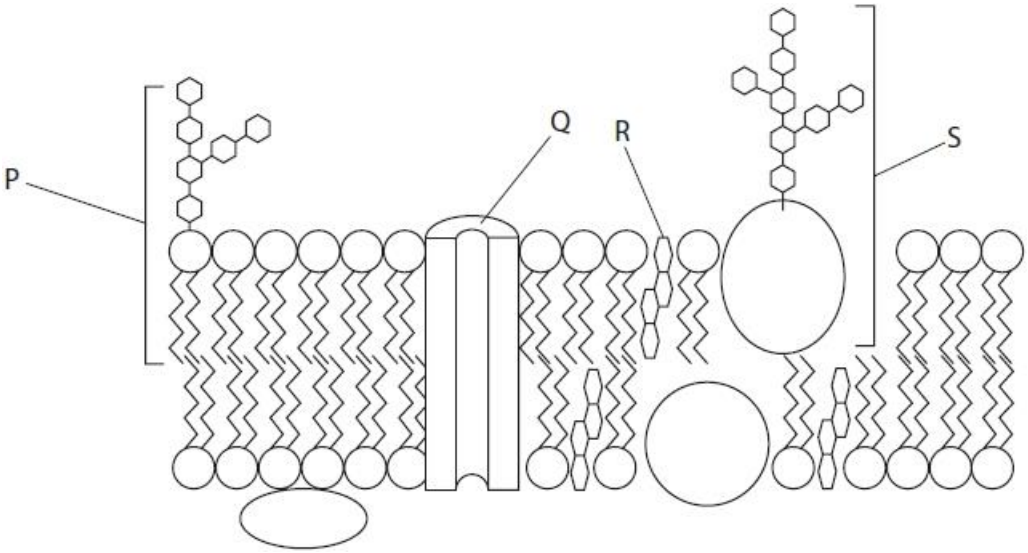
Blood type is an example of inherited variation.

Blood types A, B, AB and O are determined by a single gene.

Blood types are due to the presence or absence of antigens on the cell surface membranes of red blood cells.

These antigens are glycoproteins.

The diagram shows the structure of a cell surface membrane.



(i) Which of these labels identifies a glycoprotein?

(1)

- ☐ **A** P
- ☐ **B** Q
- ☐ **C** R
- ☐ **D** S

(ii) The alleles that produce blood type antigens A and B are codominant.

A person with the genotype I^AI^B has blood type AB.
The allele producing blood type O is recessive.
A couple have been told that the probability of having a child with blood type AB is 0.25 and the probability of blood type O is 0.25.
Deduce the genotypes and phenotypes of the parents in the table, by using a genetic diagram.

(3)

Parent	Genotype	Phenotype
1		
2		

(Total for question = 4 marks)

Q4.

The scientific article you have studied is adapted from *National Geographic*.

Use the information from the scientific article and your own knowledge to answer the following questions.

Gene drives can be used to 'force almost any genetic trait through a population' (paragraph 16).

Multiple genetic crosses were carried out between individuals homozygous for a recessive allele and individuals heterozygous for the same gene.

Describe how the outcome of these crosses would be affected if a gene drive was used with the recessive allele.

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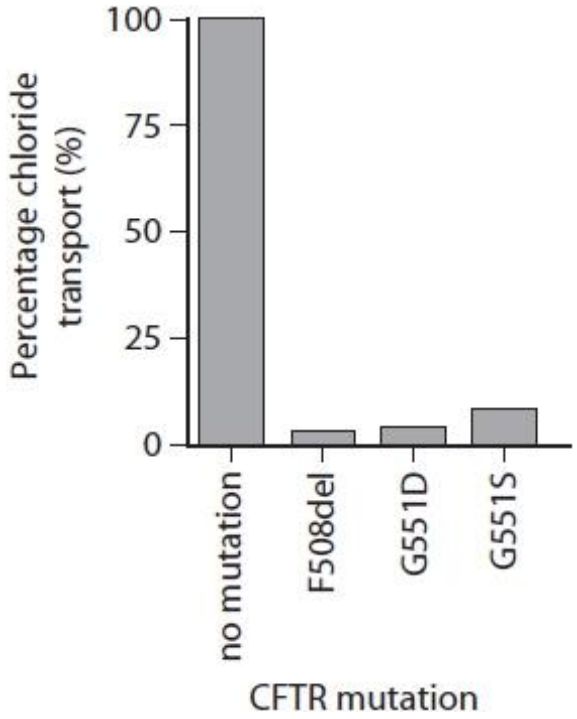
(Total for question = 3 marks)

Q5.

Mutations in genetic material such as DNA often result in the formation of new alleles.

The graph gives information about chloride transport in the human respiratory system with the normal allele for the CFTR protein and with the three mutated CFTR alleles.



The table gives information about the CFTR protein produced by cystic fibrosis (CF) sufferers with mutated alleles.

Mutation	Estimated percentage of CF sufferers who have one or more alleles with this mutation	Problem with CFTR protein channel
F508del	90	Reduced quantity / no CFTR protein
G551D	4	Reduced function
G551S	<1	Reduced function

Assess the effect that these mutations have on the human respiratory system.

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(Total for question = 6 marks)

Q6.

The food we eat contains carbohydrates, lipids and proteins.

* People with cystic fibrosis require a higher energy diet than people without cystic fibrosis. They are also more likely to develop problems in the pancreas.

Men with cystic fibrosis are less likely to be able to release sperm.

Discuss why a person with cystic fibrosis could have these symptoms.

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(Total for question = 6 marks)

Q7.

Cystic fibrosis is a condition that affects breathing.

Explain why cystic fibrosis affects the rate of oxygen uptake in the lungs.

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(Total for question = 3 marks)

Q8.

Mutations in genetic material such as DNA often result in the formation of new alleles.

More than 1500 mutations have been found for the gene that codes for the production of the CFTR channel protein.

Some of these mutations cause cystic fibrosis by affecting the production or functioning of the CFTR channel protein.

If the functioning of the CFTR channel protein is impaired, thicker mucus is produced in the lungs.

Explain why thicker mucus is produced if the functioning of the CFTR channel protein is impaired.

(2)

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(Total for question = 2 marks)

Q9.

Answer the question with a cross in the box you think is correct ☒. If you change your mind about an answer, put a line through the box ☒ and then mark your new answer with a cross ☒.

Thalassaemia is a recessive genetic disorder that affects the production of haemoglobin. It is caused by a gene mutation.

Scientists are developing methods to repair gene mutations such as the one that causes thalassaemia.

A gene mutation can be a change in a single base in the

(1)

- ☐ **A** DNA that codes for a different amino acid
- ☐ **B** DNA that codes for a different monosaccharide
- ☐ **C** RNA that codes for a different amino acid
- ☐ **D** RNA that codes for a different monosaccharide

(Total for question = 1 mark)

Q10.

Answer the question with a cross in the box you think is correct ☒. If you change your mind about an answer, put a line through the box ☒ and then mark your new answer with a cross ☒.

Cystic fibrosis can be caused by a number of different mutations in the CFTR gene.

(i) A gene contains a number of base pairs. Of the base pairs in this gene, 50% are adenine and thymine.

Q = the number of base pairs in this gene.
Which of the following shows the total number of hydrogen bonds (H bonds) present in this gene?

(1)

- ☐ **A** 2.0 × Q
- ☐ **B** 2.5 × Q
- ☐ **C** 4.0 × Q
- ☐ **D** 5.0 × Q

(ii) Explain why different mutations in the CFTR gene can lead to differences in the severity of the symptoms of cystic fibrosis.

(2)

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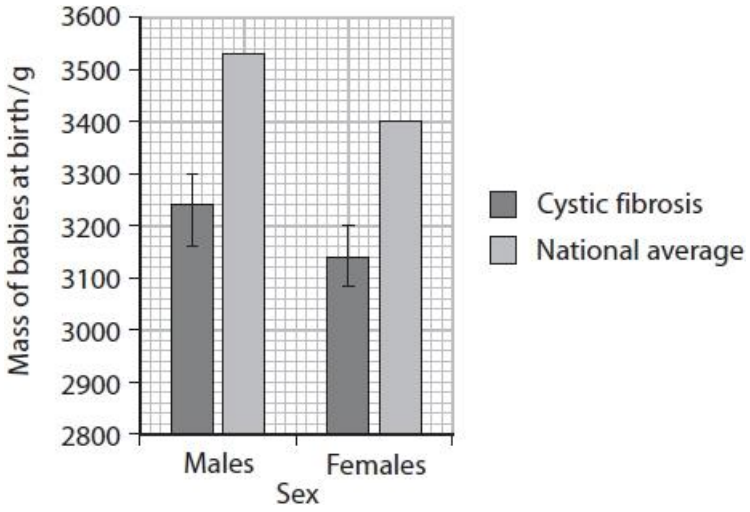
(Total for question = 3 marks)

Q11.

Answer the question with a cross in the box you think is correct ☒. If you change your mind about an answer, put a line through the box ☒ and then mark your new answer with a cross ☒.

The graph shows the mean mass of newborn babies with cystic fibrosis and of newborn babies without cystic fibrosis.



(i) How many of the following statements are correct?

- male babies always have a larger birth weight compared to female babies
- there is a significant difference between the birth weight for males and females born with cystic fibrosis
- on average female babies born with cystic fibrosis have a birth weight 260g less than the national average

(1)

☒ **A** none
☒ **B** one
☒ **C** two
☒ **D** three

(ii) Doctors give dietary supplements and digestive enzymes to children with cystic fibrosis.

Dietary supplements include carbohydrates, proteins and lipids, as well as vitamin and mineral supplements.
Explain why these children would be given dietary supplements and digestive enzymes.

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(Total for question = 6 marks)

Q12.

All cells have a cell surface membrane.

Some epithelial cells in the lungs secrete mucus. If the mucus is too 'sticky', it cannot be easily removed from the lungs.

Other epithelial cells in the lungs contain CFTR proteins in their cell surface membranes.

(i) Describe the role of the CFTR protein in ensuring that the mucus produced in the lungs has the right consistency.

(3)

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(ii) The table shows part of the gene that codes for the CFTR protein and the corresponding amino acid sequence. Each amino acid is represented by a single letter.

Part of the CFTR gene	ATTAAAGAAAATATCATCTTTGGTGTTTCCTAT										
Amino acid sequence	I	K	E	N	I	I	F	G	V	S	Y

Explain how the information in the table demonstrates the nature of the genetic code.

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(Total for question = 6 marks)

Q13.

Answer the question with a cross in the box you think is correct ☒. If you change your mind about an answer, put a line through the box ☒ and then mark your new answer with a cross ☒.

Cystic fibrosis is inherited as a recessive condition. It is caused by a mutation in the CFTR gene.

The CFTR gene codes for

(1)

- ☐ **A** a carrier protein
- ☐ **B** a channel protein
- ☐ **C** an enzyme
- ☐ **D** a glycoprotein

(Total for question = 1 mark)

Q14.

*Genetic testing can be used to identify individuals who have genetic disorders such as Batten disease.

The table shows examples of some types of genetic screening and examples of where they may be used.

Type of screening	Method	Example
Blood test to identify risk of a disease	DNA from a blood sample is examined	Identifying presence of BRCA1 and BRCA2 mutations where there is a family history of breast cancer
Blood test to identify carriers of a genetic disease	DNA from a blood sample is examined	Establishing if a person is heterozygous for a recessive condition such as cystic fibrosis (CF)
Amniocentesis	Fetal DNA from amniotic fluid is tested	Identifying genetic disorders in the fetus
Chorionic villus sampling (CVS)	Fetal DNA from placental tissue is tested	Identifying genetic disorders in the fetus
Non-invasive prenatal diagnosis (NIPD)	Analysis of fetal DNA fragments from blood samples from the mother	Identification of chromosomal disorders and a small number of single gene disorders in the fetus
Pre-implantation genetic diagnosis (PGD)	Combined with IVF to test embryo at 8-cell stage	Ensures only embryos without a genetic disorder such as CF are implanted

Assess the advantages and disadvantages of these types of screening for genetic disorders.

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(Total for question = 6 marks)

Q15.

Tay-Sachs disease is a genetic disorder.

(a) A couple without Tay-Sachs disease are expecting their second child. Their first child died from the disease.

Use a genetic diagram to determine the probability of their second child having Tay-Sachs disease.

(2)

Answer .....

(b) Tay-Sachs disease can be detected during pregnancy.

Name the prenatal test that could be used to detect Tay-Sachs disease at 11 weeks of pregnancy.

(1)

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(c) Explain why this couple may choose not to have this test.

(3)

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(Total for question = 6 marks)

Q16.

Sickle cell anaemia is a genetic disorder caused by a mutated allele for haemoglobin.

This causes one amino acid to be changed in one type of polypeptide chain in the haemoglobin protein. This affects the function of the red blood cells.

(i) An allele is a version of a gene.

State what is meant by the term gene.

(1)

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Two parents who are both heterozygous for the mutated allele are expecting a child.

Use a genetic diagram to determine the probability of this child being homozygous for the mutated allele.

(2)

Answer .....

(iii) Explain how a change of one amino acid could lead to a change in the structure and properties of the haemoglobin protein.

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(Total for question = 7 marks)

Q17.

Answer the question with a cross in the box you think is correct ☒. If you change your mind about an answer, put a line through the box ☒ and then mark your new answer with a cross ☒.

Batten disease is a rare, inherited disorder of the nervous system. It usually begins in childhood. It is a recessive disorder.

(i) Parents without Batten disease have a child with Batten disease.

Which of the following describes the genotype of the parents?

(1)

- ☐ **A** bb and Bb
- ☐ **B** Bb and Bb
- ☐ **C** BB and BB
- ☐ **D** BB and bb

(ii) Draw a genetic diagram to show the probability of their future children developing Batten disease.

(2)

(Total for question = 3 marks)

Q18.

Batten disease is a rare, inherited disorder of the nervous system. It usually begins in childhood. It is a recessive disorder.

Explain what is meant by an inherited recessive disorder.

(2)

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(Total for question = 2 marks)

Q19.

Answer the question with a cross in the box you think is correct ☒. If you change your mind about an answer, put a line through the box ☒ and then mark your new answer with a cross ☒.

The phenotype of organisms is affected by their genotype.

Cattle have different patterns and colours in their coats as a result of their genotype.

The image shows the genotype and phenotype of three offspring from the same parents.

pure red (RR) | roan (RW) | pure white (WW)

(Source from: http://wps.pearsoned.com.au/wps/media/objects/8476/8680015/_images_/ch3c.jpg)

(i) Name the type of inheritance shown in this example.

(1)

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(ii) What is the probability of the next offspring of the same parents being roan?

(1)

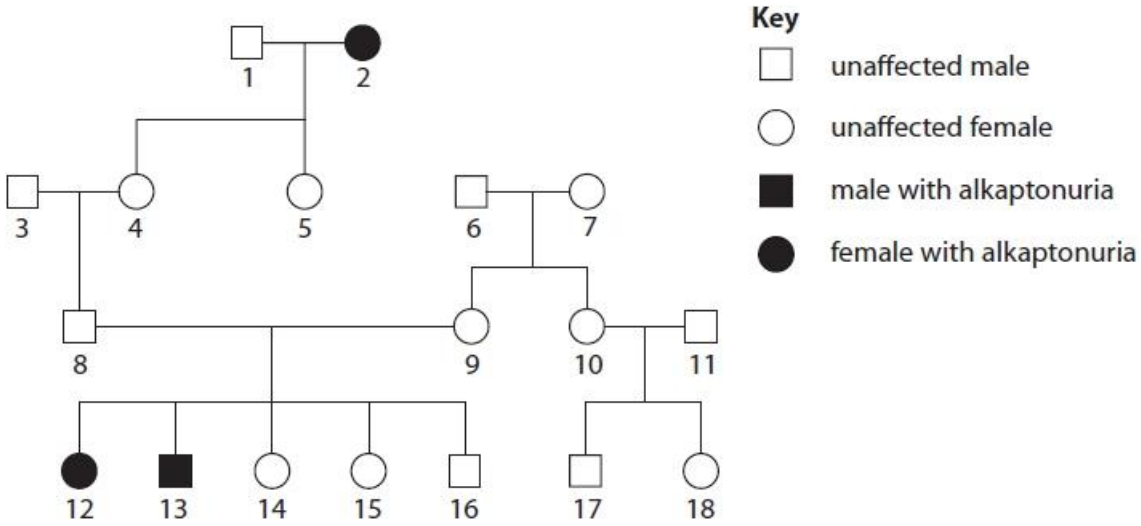
- ☒ **A** 25%
- ☒ **B** 50%
- ☒ **C** 75%
- ☒ **D** 100%

(Total for question = 2 marks)

Q20.

Alkaptonuria is a rare condition where the body cannot break down the amino acids phenylalanine and tyrosine.

The pedigree diagram shows part of a family tree in which alkaptonuria is an inherited condition.



(i) State and justify whether alkaptonuria is caused by a recessive or a dominant allele.

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(ii) Individual 12 is pregnant and wants to know if her baby has alkaptonuria.
State and justify a suitable method of collecting cells for prenatal testing.

(3)

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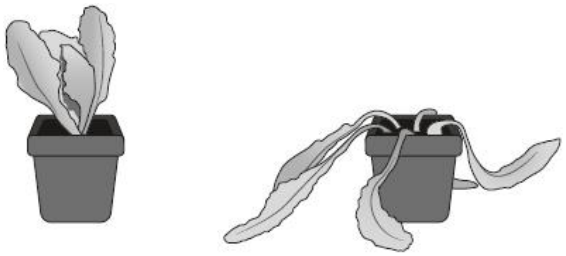
(Total for question = 7 marks)

Q21.

Lettuce plants usually grow upright. This is the 'upright' phenotype.

In one variety of lettuce the stem of the lettuce grows along the ground. This is the 'weary' phenotype.

These two phenotypes are shown in the diagram.



Upright Weary

Inheritance of the weary phenotype has been investigated.

Scientists crossed weary lettuce plants with upright lettuce plants.
The F_1 generation produced from this cross were all upright.

In the second cross, two of the F_1 lettuce plants were crossed with each other to produce the F_2 generation.

The phenotypes of the F_2 generation and the results of a statistical test are shown in the table.

Number of offspring with weary phenotype	Number of offspring with upright phenotype	Chi-squared (χ^2)
159	414	2.31

Degrees of freedom	Probability		
	0.01	0.05	0.1
1	2.71	3.84	6.64
2	4.61	5.99	9.21
3	6.25	7.82	11.35
4	7.78	9.49	13.28

Justify the conclusion that the weary phenotype was inherited as a recessive trait.

(3)

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(Total for question = 3 marks)

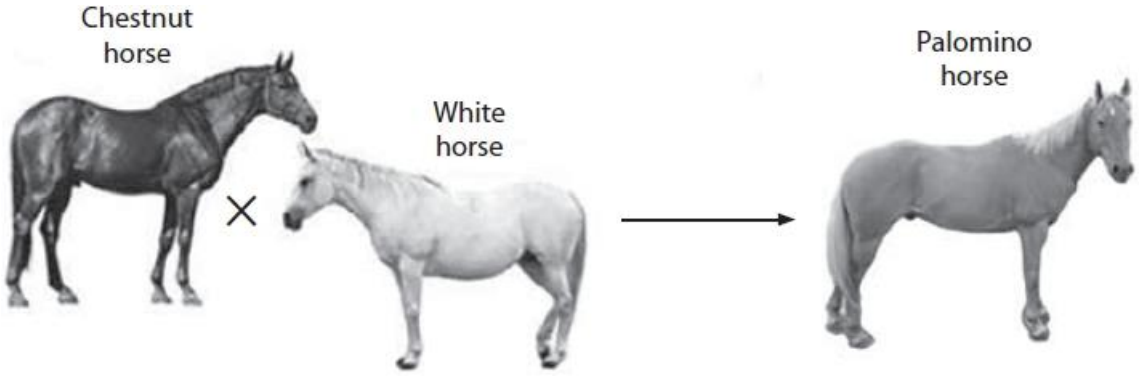
Q22.

The phenotype of organisms is affected by genotype.

Chestnut horses are homozygous for the allele H^C. White horses are homozygous for the allele H^W.

If a chestnut horse is mated with a white horse, the offspring will be palomino.

Palomino horses have coats with a colour intermediate between chestnut and white.



(i) State what is meant by the term **allele**.

(2)

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(ii) Explain why the offspring have the palomino coat colour.

(3)

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(Total for question = 5 marks)

Mark Scheme

Q1.

Question Number	Answer	Additional guidance	Mark
	An answer that makes reference to the following • correct genetic diagram used to determine genotypesof offspring (1) • correct probability 0.5 linked to correct genotypes ofoffspring (1)	e.g. S s s Ss ss s Ss ss Ss and ss ALLOW 50% / ½ / 1 in 2	**(2)**

Q2.

Question Number	Answer	Additional guidance	Mark
	An answer that makes reference to the following: • 0.5 probability for being { same sex / female} (1) • person 1 is heterozygous for MPS 1 (1) • (therefore) person 2 has a 0.75 probability of having same phenotype as person 1 for MPS 1(1) • therefore probability of being female and nothaving MPS 1 will be 0.375 (1)	ALLOW 50% for 0.5 ALLOW detail of proof of phenotype of person 1 e.g. does not show condition therefore has to have one dominant allele but (at least) one daughter has condition so received a recessive allele from person 1 ALLOW carrier for heterozygous ALLOW 75% for 0.75 ALLOW $^{3}/_{8}$ or 37.5% for 0.375	**(4)**

Q3.

Question Number	Answer	Additional Guidance	Mark
(i)	D–S *A is incorrect because it is a glycolipid* *B is incorrect because it is an intrinsic protein* *C is incorrect because it is cholesterol*		**(1)**

Question Number	Answer	Additional Guidance	Mark
(ii)	An answer that makes reference to the following: • genotype of one parent has one A allele (and no B) (1) • genotype of other parent has one B allele (and no A allele) (1) • correct phenotypes identified – (blood group) A for one parent and B for the other parent (1)		**(3)**

Q4.

Question number	Answer	Additional guidance	Mark
	A description that makes reference to the following: • without a gene drive the expected outcome wouldbe 50% heterozygous and 50% homozygous recessive offspring (1) • with a gene drive the proportion of homozygous recessive offspring would increase (1) • the stronger the gene drive the greater the proportion of homozygous recessive (1)	ALLOW a genetic cross diagram toshow this	Choose an item. (3)

Q5.

Question Number	Answer	Mark
	Answers will be credited according to candidate's knowledge and understanding of the material in relation to the qualities and skills outlined in the generic mark scheme. The indicative content below is not prescriptive and candidates are not required to include all the material which is indicated as relevant. Additional content included in the response must be scientific and relevant. Basic information • cilia struggle to move mucus out of lungs • increased risk of lung infections • 100% chloride ion transport without mutation • all three mutations reduce chloride transport • F508del results in lower chloride transport than {the other mutations / G551D / G551S } • G551S mutation is less severe than G551D Evidence for linkages made • F508del mutation results in { no / fewer } CFTR protein channels being produced • G551D and G551S have higher chloride transport (than F508del) because { the correct number of protein channels are produced / CFTR protein present but function reduced } • {no/fewer} CFTR protein channels results in less transport of chloride ions • effects of thicker mucus on gas exchange in the respiratory system explained e.g. increased diffusion distance, reduced surface area, reduced concentration gradient Evidence for sustained scientific reasoning • with the G551D and G551S mutations the CFTR protein channels have an incorrect shape • incorrect shape of CFTR protein results in reduced function (of transporting chloride ions) • a different part of the protein structure is affected in G551S which interferes less in the transport of chloride ions • mutation may affect the tertiary structure of the CFTR protein e.g. the positioning of hydrophilic parts of the protein channel	**(6)**

			Additional guidance
Level 0	0	No awardable content	
Level 1	1-2	An answer may be attempted but with limited interpretation or analysis of the scientific information with a focus on mainly just one piece of scientific information. The explanation will contain basic information with some attempt made to link knowledge and understanding to the given context.	Basic description of the effects of the mutations on CFTR protein production. E.g. comparisons between mutations and no mutations on CFTR production Effects of CFTR protein problems on mucus described
Level 2	3-4	An answer will be given with occasional evidence of analysis, interpretation and/or evaluation of both pieces of scientific information. The explanation shows some linkages and lines of scientific reasoning with some structure.	Explanation of how the different types of mutation will have different effects on CFTR and chloride ion transport. Effects of CFTR protein problems on respiratory system described
Level 3	5-6	An answer is made which is supported throughout by sustained application of relevant evidence of analysis, interpretation and/or evaluation of both pieces of scientific information. The explanation shows a well-developed and sustained line of scientific reasoning which is clear and logically structured.	Explanation of effects of mutations related to shape of protein produced. Reasons for reduced function of CFTR protein related to G551S and G551D mutations.

Q6.

Question Number	Answer
*	Answers will be credited according to candidate's knowledge and understanding of the material in relation to the qualities and skills outlined in the generic mark scheme. The indicative content below is not prescriptive and candidates are not required to include all the material which is indicated as relevant. Additional content included in the response must be scientific and relevant. • mucus {thicker / stickier} than normal • (pancreatic) enzymes cannot enter intestine because pancreatic duct blocked with mucus • (high energy diet required because) digestion is less efficient • pancreatic enzymes trapped behind mucus damage pancreatic cells such as those that produce insulin • cysts form in pancreas • sperm cannot leave the testes because {sperm duct / vas deferens / tubes } blocked with mucus • sperm duct / vas deferens absent therefore sperm cannot pass through • gene mutation • (causing a) non-functioning CFTR protein channel • chloride ions cannot move out of epithelial cells • accumulation of sodium and chloride ions in the cells {causing water to move out of mucus by osmosis / preventing water moving into mucus} **(6)**

Level 0	0	No awardable content	
Level 1	1-2	Demonstrates isolated elements of biological knowledge and understanding to the given context with generalised comments made. Vague statements related to consequences are made with limited linkage to a range of scientific ideas, processes, techniques and procedures. The discussion will contain basic information with some attempt made to link knowledge and understanding to the given context.	Answers discuss at least one reason with limited reference to relevant science.
Level 2	3-4	Demonstrates adequate knowledge and understanding by selecting and applying some relevant biological facts/concepts. Consequences are discussed which are occasionally supported through linkage to a range of scientific ideas, processes, techniques and procedures. The discussion shows some linkages and lines of scientific reasoning with some structure.	Reference is made to sticky/thick mucus. Two out of the three statements are discussed with reference to relevant science.
Level 3	5-6	Demonstrates comprehensive knowledge and understanding by selecting and applying relevant knowledge of biological facts/concepts. Consequences are discussed which are supported throughout by sustained linkage to a range of scientific ideas, processes, techniques or procedures. The discussion shows a well-developed and sustained line of scientific reasoning which is clear and logically structured.	Reference is made to {gene mutation / non-functioning protein channel} and {sticky/thick} mucus. Correct and detailed science is used to explain all 3 statements in a clear and logical way

Q7.

Question number	Answer	Additional guidance	Mark
	An explanation that makes reference to three of the following: • { thick / sticky / viscous } mucus (1) • (accumulation of mucus) which cannot be moved by cilia (1) • restricting air flow through { bronchioles / bronchi } (1) • {increases diffusion distance / reduces surface area for gas exchange } in the alveoli (1)	IGNORE 'airways' ALLOW narrowing of bronchioles	(3)

Q8.

Question Number	Answer	Additional Guidance	Mark
	An explanation that makes reference to two of the following: • chloride ions cannot {leave the cell / enter mucus} (through CFTR protein channel) (1) • sodium ions do not move { out of the cells / into the mucus } (1) • therefore water {moves into cells / moves out of mucus} by osmosis (1)	ALLOW water does not move into mucus by osmosis	**(2)**

Q9.

Question Number	Answer	Mark
	The only correct answer is A - DNA which codes for a different amino acid *B is incorrect because DNA does not code for monosaccharides* *C is incorrect because the change is not in RNA* *D is incorrect because the change is not in RNA and it does not code for monosaccharides*	**(1)**

Q10.

Question Number	Answer	Mark
(i)	The only correct answer is B - 2.5 x Q *The only correct answer is B because 50% of the base pairs are A-T with 2 hydrogen bonds and 50% C-G with 3 hydrogen bonds making a mean of 2.5 x Q* *A is incorrect because 2.0 x Q is only correct if all base pairs were A-T* *C is incorrect because it assumes that each base in a pair has 4 hydrogen bonds and all the base pairs are A-T* *D is incorrect because it assumes that each base in the A-T base pair has 4 hydrogen bonds and each base in the G-C has 6 hydrogen bonds*	**(1)**

Question Number	Answer	Additional Guidance	Mark
(ii)	An explanation that makes reference to two of the following: • (different mutations) will have different effects on the protein produced (1) • chloride ion transport affected by the extent of changes to the (CFTR) protein (1) • varying the { stickiness / thickness } of the mucus (1)	ALLOW absence of protein / different { folding / tertiary structure / shape } of the protein ALLOW faulty CFTR protein has less impact on chloride ion transport than a missing CFTR protein	**(2)**

Q11.

Question Number	Answer	Mark
(i)	The only correct answer is – B - one A is incorrect because the third statement is correct C is incorrect because statements one and two are incorrect D is incorrect because statements one and two are incorrect	(1)

Question Number	Answer	Additional guidance	Mark
(ii)	An explanation which makes reference to five of the following: • cystic fibrosis causes the production of {thicker / stickier} mucus (1) • which blocks the pancreatic duct / prevent pancreas enzymes reaching intestine (1) • {reduces / prevents} enzymes digesting {carbohydrates / lipids / proteins} (in intestines) (1) • (resulting in) reduced {absorption / concentration} of products of digestion into the {blood / lymph} (1) • linkage of reduced {amino acids / vitamins / minerals} to slower growth rate (1) • {dietary supplements / digestive enzymes} would increase growth rate (1)	ALLOW converse for given digestive enzymes for mp3+4 ALLOW reduced digestion of food by enzymes e.g. amino acids / glucose / fatty acids / glycerol / vitamins / minerals	(5)

Q12.

Question Number	Answer	Additional Guidance	Mark
(i)	A description that makes reference to three of the following: • chloride ions leave cells (through the CFTR channel protein) (1) • sodium ions leave the cells (following the chloride ions) (1) • increasing the solute concentration in the mucus (1) • water moves out of the cells by osmosis (into the mucus) (1)	NOT active transport of chloride ions ALLOW chloride ions move into the mucus ALLOW NaCl, Na^+ or Cl^- instead of solute ALLOW description of osmosis	3

Question Number	Answer	Additional Guidance	Mark
(ii)	An explanation that makes reference to the following: • (triplet code) is shown by three bases coding for an amino acid (1) • non-overlapping code e.g. ATT codes for amino acid I and then AAA code for amino acid K (1) • degenerate code as both ATT and ATC code for amino acid I (1)		3

Q13.

Question Number	Answer	Mark
	The only correct answer is – B - a channel protein A is incorrect because it is not a carrier protein C is incorrect because it is not an enzyme D is incorrect because it is not a glycoprotein	(1)

Q14.

Question Number	Answer
*	Answers will be credited according to candidate's deployment of knowledge and understanding of the material in relation to the qualities and skills outlined in the generic mark scheme. The indicative content below is not prescriptive and candidates are not required to include all the material which is indicated as relevant. Additional content included in the response must be scientific and relevant. <u>Indicative content</u> Adult screening advantages and disadvantages • Identifies risk of developing a particular disease in the future so choices can be made e.g. extra screening for breast cancer or preventative mastectomy/screening and lifestyle changes for some types of CVD • Identification of carriers so choices can be made about family planning – both partners tested, risk can be identified and have prenatal screening • May not want to know if you have a high likelihood of developing a disease, if one person is tested it may give other family members information they would chose not to know, may potentially affect life insurance

	Prenatal screening advantages and disadvantages • Amniocentesis – prepares parents for child with disease/gives choice of abortion • Chorionic villus sampling – as amniocentesis, carried out earlier in pregnancy • Some of the conditions tested for are very unpleasant and may be life limiting • NIPD non-invasive, less traumatic procedure, no increased risk of miscarriage • PGD only implant healthy embryos, do not have to make decision about abortion • Both amniocentesis and CVS carry increased risk of miscarriage, especially CVS (although it can be carried out earlier in the pregnancy) • Can't cure the disease, only choice is to have an abortion-not acceptable to everyone • For conditions such as CF, where there is more than one possible mutation, test is only for most common mutations so there may be false negatives • NIPD currently only available for chromosome disorders such as Down's syndrome • PGD involves IVF, which can be emotionally traumatic and only has about 30% success rate • All pre-natal screening has a risk of false positives with abortion of a healthy fetus. • Procedures involving IVF can be regarded as unethical because many embryos are discarded • Invasive nature of some of the tests

Level	Mark	Descriptor	Additional Guidance
0	Mark	No awardable content	
1	1-2	Limited scientific judgement made with a focus on one side of the argument only. A conclusion may be attempted, demonstrating isolated elements of biological knowledge and understanding but with limited evidence to support the judgement being made.	Only considered one benefit or one risk without further explanation beyond a brief description. A generalised discussion without focusing on the details of specific types of screening
2	3-4	A scientific judgement is made through the application of relevant evidence to both sides of the argument. A conclusion is made, demonstrating linkages to elements of biological knowledge and understanding, with occasional evidence to support the judgement being made.	Considers at least two types of screening One advantage and one disadvantage of each type of screening discussed.
3	5-6	A scientific judgement is made, which is supported throughout by sustained application of relevant evidence from the analysis and interpretation of the scientific information. A conclusion is made, demonstrating sustained linkages to biological knowledge and understanding with evidence to support the judgement being made.	Advantages and disadvantages of blood tests and pre-natal tests discussed fully. Discussion of blood tests to identify adults with genetic disorders. Conclusion or judgement made, e.g. identifying genetic disorders by blood tests in adults is better as the disadvantages have less impact than disadvantages of genetic testing on embryos / fetuses.

Q15.

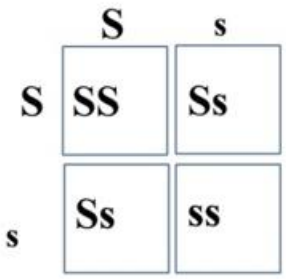
Question Number	Answer	Additional Guidance	Mark
(a)	• correct genetic diagram with reference to parental and offspring genotypes (1) • correct probability (1)	Allow correct gametes and offspring genotypes e.g. T and t for gametes, offspring TT, Tt and tt 0.25/ 25% / $\frac{1}{4}$ Do not accept 1:4 or 1:3	(2)

Question Number	Answer	Additional Guidance	Mark
(b)	chorionic villus sampling	Accept CVS Accept phonetic spelling Do not accept chronic villus sampling	(1)

Question Number	Answer	Additional Guidance	Mark
(c)	An explanation that makes reference to three of the following: • test result may be inaccurate (1) • (increased) risk of miscarriage (due to the procedure) (1) • false positive may lead to termination of healthy fetus (1) • { prenatal testing / abortion } against values or beliefs of the parents (1)	Ignore reference to amniocentesis Allow false positives / false negatives Allow spontaneous abortion Allow 'parents do not want to know'	(3)

Q16.

Question Number	Answer	Additional Guidance	Mark
(i)	An answer which makes reference to the following: • sequence of {bases / nucleotides} in DNA coding for a {sequence of amino acids / polypeptide / protein}	ALLOW section of DNA coding for a {sequence of amino acids / polypeptide / protein}	(1)

Question Number	Answer	Additional Guidance	Mark
(ii)	• correct genetic diagram with reference to parental and offspring genotypes (1)	ALLOW correct gametes and offspring genotypes e.g. S and s for gametes, offspring SS, Ss and ss (or other letters)  	
	• correct probability (1)	0.25 / 25% / ¼ IGNORE ratios	(2)

Question Number	Answer	Additional Guidance	Mark
(iii)	An explanation that makes reference to the following: • different {sequence of amino acids / primary structure} (1) • (a different amino acid will have a) different R group (1) • (therefore) {secondary / tertiary / quaternary} structure will change (1) • (due to a) change in a named bond (holding molecule in its three-dimensional shape) (1) • (haemoglobin) may not bond to oxygen (1)	ALLOW different polypeptide chain i.e. hydrogen bonds, disulfide bridges, ionic bonds DO NOT ALLOW peptide bonds ALLOW may not bond to haem group ALLOW may not carry oxygen	**(4)**

Q17.

Question Number	Answer	Mark
(i)	***The only correct answer is B*** Bb and Bb ***A*** *is not correct because the parent who is bb would have Batten disease* ***C*** *is not correct because neither parent has the b allele* ***D*** *is not correct because the parent who is bb would have Batten disease and the parent who is BB would not have the b allele*	**(1)**

Question Number	Answer	Additional guidance	Mark
(ii)	An answer that makes reference to the following: • correct genetic diagram with reference to offspring genotypes (1) • correct probability of inheriting Batten disease (1)	e.g. BB, Bb, Bb and bb 1 in 4 / 25% / 0.25 IGNORE ratios	**(2)**

Q18.

Question Number	Answer	Additional guidance	Mark
	An explanation that makes reference to the following: • (a recessive disorder is one) caused by a faulty allele (1) • that is only expressed in the { homozygous condition / absence of a normal allele } (1)	ALLOW faulty gene ALLOW only expressed if genotype is { homozygous recessive / bb } or if two recessive alleles are inherited	**(2)**

Q19.

Question	Answer	Additional guidance	Mark
(i)	• incomplete dominance (1)	ALLOW co-dominance	(1)

Question Number	Answer	Mark
(ii)	The only correct answer is – B - 50% A is incorrect because 25% is the probability for either RR or WW C is incorrect because two heterozygous parents would not result in 75% RW D is incorrect because two heterozygous parents would not result in 100% RW	(1)

Q20.

Question Number	Answer	Additional guidance	Mark
(i)	An answer which makes reference to the following: • recessive (1) • two recessive alleles are needed to have condition (1) • individual(s) {8 / 9} do not have alkaptonuria but some of their children do (1) • therefore individuals 8 and 9 must be heterozygous (1)	ALLOW carriers	(4)

Question Number	Answer	Additional guidance	Mark
(ii)	An answer which makes reference to the following: <u>Either</u> • chorionic villus sampling / CVS (1) • {cells/ tissue} taken from {placenta / chorionic villus} between 10-14 weeks of pregnancy (1) • benefit of earlier diagnosis (1) <u>Or</u> • amniocentesis (1) • amniotic fluid containing cells collected between 14-20 weeks of pregnancy (1) • lower risk of miscarriage (1)		(3)

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Q21.

Question Number	Answer	Additional guidance	Mark
	An answer the makes reference to three of the following: • no offspring from the cross between weary and upright lettuce had the weary phenotype (1) • the ratio of weary to upright lettuce in the F_2 generation was 1: 3 (1) • the chi-squared test value was below the critical value (1) • result not statistically different from expected result (1)	ALLOW none of the F_1 generation had the weary phenotype / all the F_1 generation were upright ALLOW {25% / ¼ / 27.7%} of the F_2 generation were weary lettuce ALLOW less than a critical value of 3.84 IGNORE degrees of freedom or incorrect cv ALLOW the null hypothesis can be accepted	(3)

Q22.

Question Number	Answer	Additional Guidance	Mark
(i)	An answer which makes reference to the following: • alternative form of a gene (1) • found at the same locus (on a chromosome) (1)	ALLOW different version of a gene ALLOW same place on a chromosome	(2)

Question Number	Answer	Additional Guidance	Mark
(ii)	An explanation which makes reference to the following: • (palomino / offspring) is heterozygous (1) • therefore { incomplete dominance / co-dominance } occurs (1) • because both alleles are expressed (1)	ALLOW palomino horses have the genotype $H^C H^W$ or correct genetic diagram to show this ALLOW alleles are co-dominant	(3)