1. Thirty-three human blood group systems are known to exist. Two of these are the ABO blood group system and the Hh blood group system.

Explain why a person whose blood group is AB expresses both A and B antigens on the surface of their red blood cells.

---

[2]

2. Some varieties of maize plants have smooth kernels (seeds), whereas others have wrinkled kernels. This is a genetic trait.

Varieties with smooth kernels are rich in starch and useful for making flour.

A farmer has been given some smooth seeds all of the same unknown genotype. The farmer carries out a cross-breeding experiment using these seeds and some known to be heterozygous for this trait.

The results are shown in Table 4.1.

F_1 phenotype	Observed results	Expected results
Smooth	547	
Wrinkled	185	
Total	732	

Table 4.1

The χ^2 statistic is calculated in the following way:

$$\chi^2 = \sum \frac{(\text{observed} - \text{expected})^2}{\text{expected}}$$

(i) Calculate the value of χ^2 for the above data. Show your working.

Answer _____ [2]

(ii) **Table 4.2** shows a critical values table.

Degrees of freedom	probability, p			
	0.90	0.50	0.10	0.05
1	0.016	0.455	2.71	3.84
2	0.211	1.386	4.61	5.99
3	0.584	2.366	6.25	7.81
4	1.064	3.357	7.78	9.49

Table 4.2

Using your calculated value of χ^2 and Table 4.2 what conclusions should you make about the significance of

the difference between the observed and expected results?

---

[2]

(iii) Describe the genotype of the seed the farmer was given. Give a reason for your answer.

---

[2]

3(a). A successful organ transplant usually depends on a good genetic match between the organ donor and the recipient to avoid the transplanted organ being rejected.

Complete the following paragraph about the major histocompatibility (**MHC**) system by inserting the most appropriate term.

There are six major genes controlling the MHC complex located on chromosome 6. Each gene codes for a

_____, which is expressed on the cell surface membrane and acts as an

_____ Each of the six genes has many different alleles.

The gene _____ are close together on chromosome 6, making it unlikely that

_____ will occur between them during prophase 1 of _____

This means that one complete set of alleles or **haplotype** will be inherited from each parent. The MHC alleles

are said to be _____, as the alleles from both haplotypes are expressed in the phenotype.

[6]

(b). A transplant is more likely to be successful if MHC haplotypes and the ABO blood group of the donor and the recipient are matched.

The genes controlling the ABO blood group are located on chromosome 9.

Explain how the children of parents who are heterozygous for blood group A could have identical MHC haplotypes but different blood groups.

---

[3]

4(a). Choroideremia is an inherited sex-linked recessive condition that results in degeneration of the choroid layer in the eye. This condition leads to a gradual breakdown of the retina and eventual blindness.

Using the normal conventions for constructing genetic diagrams and the letters **E** and / or **e**, choose appropriate symbols to represent:

(i) the allele for choroideremia

---

[1]

(ii) the possible genotype(s) of a person who develops choroideremia

---

[1]

(iii) the genotype of a carrier of choroideremia.

---

[1]

(b). The gene involved in choroideremia codes for a protein called REP–1.

Many of the gene mutations that cause choroideremia result in the formation of a protein that is much smaller than normal REP–1. This smaller protein is known as a **truncated protein**.

(i) State the **organelle** in the cell where the following occurs:

a complementary RNA copy of the gene is synthesised ---

the REP-1 protein is synthesised ---

[2]

(ii) Suggest **how** a mutation in the REP-1 gene could lead to the formation of a truncated protein.

---

[2]

(c). A phase 1 clinical trial was carried out using gene therapy to treat choroideremia. An outline of the method used is given below.

- A complementary DNA (cDNA) copy of the REP–1 gene was inserted into a viral vector.
- The retina of the patient was detached to expose the choroid layer.
- A fine needle was used to inject the virus into the choroid layer.

(i) Name the enzyme used to create a cDNA copy of the REP–1 gene.

---

[1]

(ii) What type of gene therapy has been used in the trial?

---

[1]

(iii) Discuss the reasons why genetic diseases such as choroideremia are good choices for treatment using gene therapy.

---

[3]

5. Human genetic diseases, such as sickle cell anaemia and cystic fibrosis, are caused when mutated forms of a gene are inherited.

The study of how these alleles are distributed in human populations can be an indication of how a human population has evolved over time.

State **two** genetic diseases, other than sickle cell anaemia and cystic fibrosis, which are caused by the inheritance of mutant alleles.

1

2

---

[2]

6(a). Down's syndrome (DS) is a genetic condition that occurs in approximately 1 in every 800 births.

Fig. 2.1a shows a karyotype from a male with a rare form of DS. In this karyotype, there is an additional piece of genetic material attached to one copy of chromosome 14.

A larger diagram of both copies of chromosome 14 is shown in **Fig. 2.1b**.

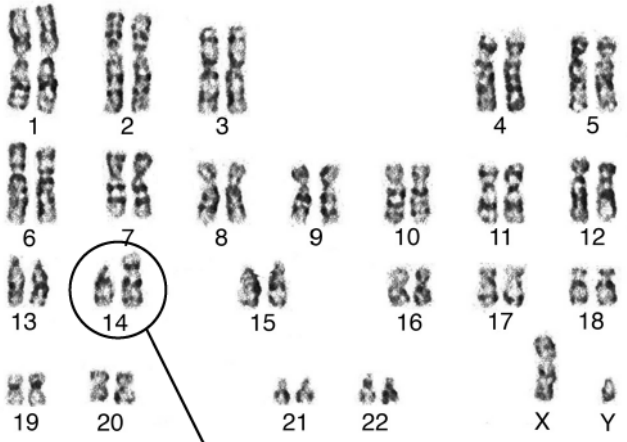


Fig. 2.1a

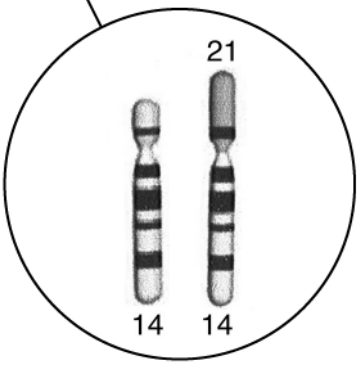


Fig. 2.1b

- Describe how the appearance of the karyotype shown in Fig. 2.1a differs from that of the more common form of DS.
- Suggest **when** and **how** the transfer of extra genetic material to chromosome 14 occurred, and what events led to cells with this karyotype being formed.

In your answer, you should include a comparison of the karyotypes and describe the events during and after meiosis that led to the formation of the karyotype in ***Fig. 2.1a****.*

[7]

(b). The National Cytogenic Register for DS was set up in the United Kingdom in 1989.

This register holds anonymous data on families for over 26 000 cases of DS. These cases were diagnosed either antenatally or postnatally.

Fig. 2.2 shows the changes in the number of DS cases diagnosed between 1990 and 2005. It also shows the changes in the number of live births of children with DS for the same period.

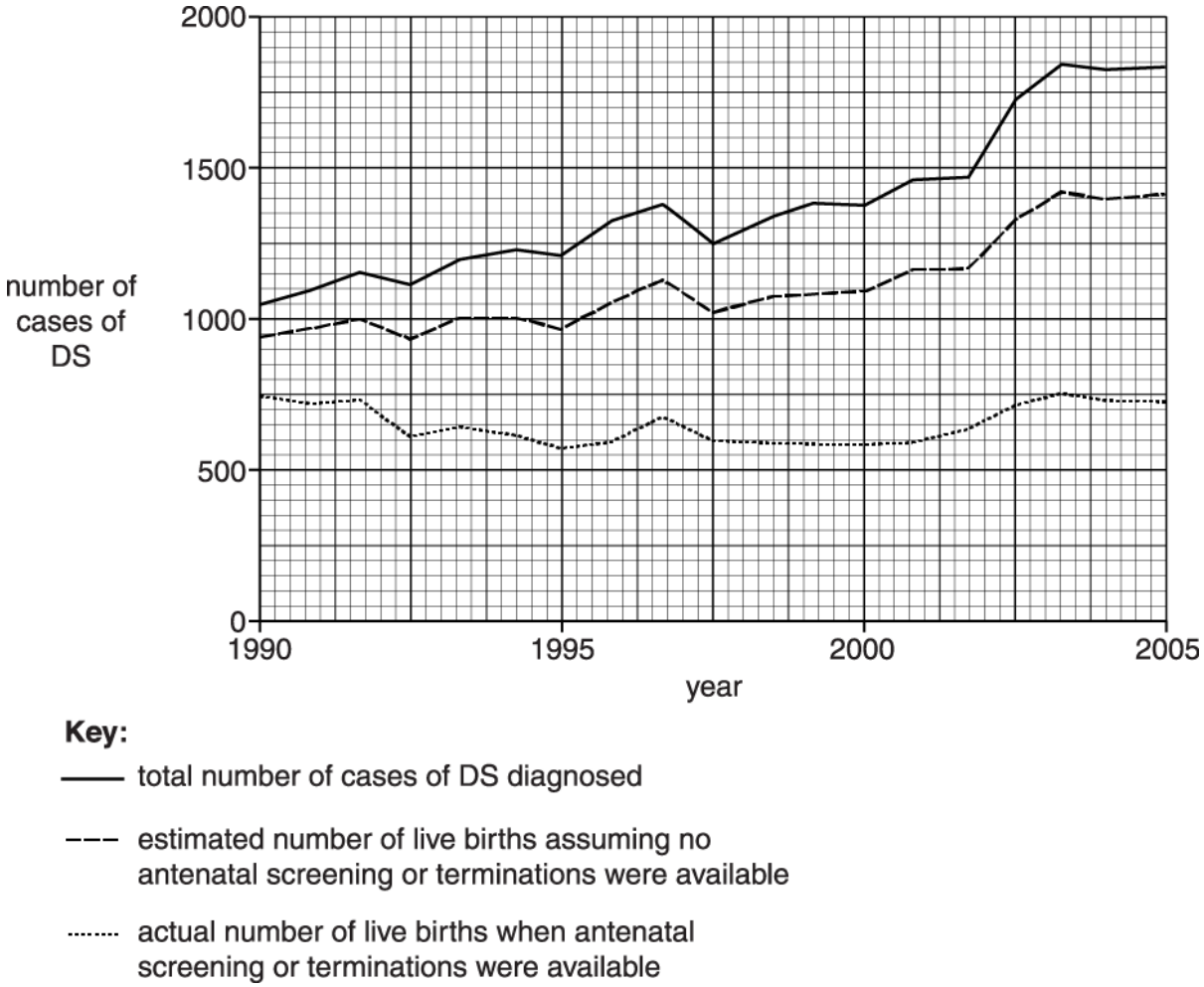


Fig. 2.2

(i) Suggest why the data on the National Cytogenic Register for DS are held anonymously.

---

[1]

(ii) Using the information in Fig. 2.2, suggest **why** the number of cases of DS **diagnosed** between 1990 and 2005 has changed.

[2]

(iii) Suggest what factor the investigators took into account in estimating the number of children with DS who would have been **born**, assuming that no antenatal screening or terminations were available.

[1]

7. The appearance of blood samples viewed under a microscope can be used to diagnose a number of different conditions.

Fig. 7.1 shows a blood sample. An abnormal cell known as a **schistocyte** is labelled. Schistocytes are formed from fragments of red blood cells.

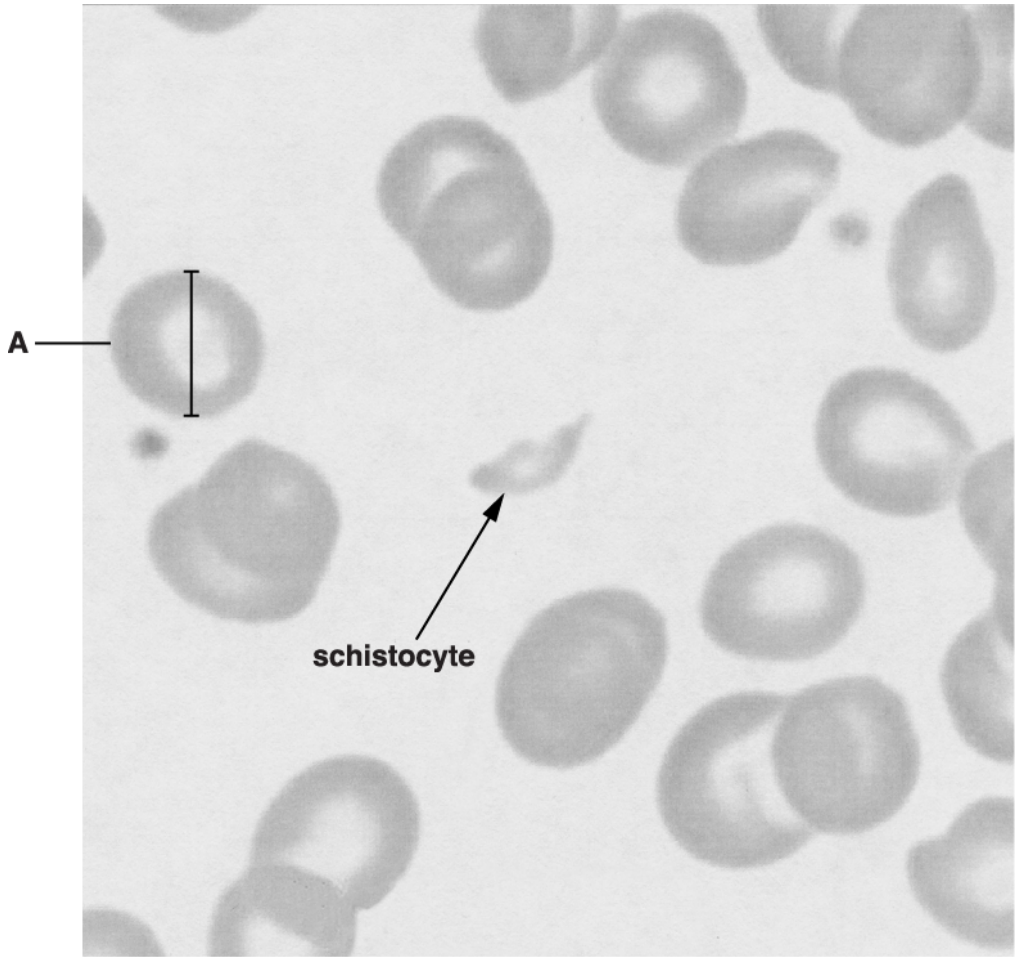


Fig. 7.1

(i) Use the information in Fig. 7.1 to calculate the size of cell **A**.

Show your working. Give your answer to the nearest whole number.

Answer = _____ μm

[2]

(ii) When activated platelets bind to the endothelium of small arterioles and capillaries, schistocytes are formed.

Suggest why the binding of activated platelets leads to the formation of schistocytes.

_____

_____ [1]

(iii) State one **genetic** disease that could be diagnosed by examination of a blood smear.

_____ [1]

8. Haemophilia A is a disease caused by a mutation in the gene coding for Factor VIII.

Factor VIII is involved in the blood clotting cascade.

(i) Scientists have found different types of mutation that can cause haemophilia A.

One type of mutation is a deletion of **two** bases. Suggest how this mutation causes the Factor VIII protein to lose its biological function.

---

[2]

(ii) Explain why haemophilia A is more common in males than in females.

---

[2]

(iii) A male with haemophilia A and a female who carries the disease have a child.

Complete the Punnett square below to determine the probability that the child will have haemophilia A. Use **H/h** to represent the alleles coding for Factor VIII.

male

		.....	.....
female	.....	.....	.....
	.....	.....	.....

probability _____ **[3]**

9. The Galapagos Islands is an ecosystem of exceptional biological interest.

The lava cactus, *Brachycereus nesioticus*, is found only in the Galapagos Islands. It speciated rapidly from a very few individuals of a parent species, perhaps only two. These individuals were carried on currents from the mainland of South America.

There is a gene that enables the mainland *Brachycereus* species to obtain water from damp mists in the atmosphere:

- let Q be the normal allele, allowing the cactus to obtain water from damp mists
- let q be a rare recessive allele that, when homozygous, could allow the cactus to obtain water from salty sea spray.

(i) Consider a cross between two heterozygous individuals.

What is the **theoretical** percentage of the offspring from these two individuals that would be able to obtain water from sea spray?

Use the space below for any working.

Answer = _____ % **[1]**

(ii) *B. nesioticus* colonises bare rock at the edge of the Galapagos Islands.

Explain how individuals homozygous for the q allele would soon come to dominate the gene pool.

_____

_____

_____

_____

_____

_____

_____

[4]

10(a) The gene, *HBB*, codes for the beta polypeptide in haemoglobin.

.

A person with sickle cell anaemia has a mutation in *HBB*. This causes a change to the sixth amino acid in the beta polypeptide.

Describe the type of DNA mutation that causes the amino acid change in the beta polypeptide.

---

[1]

(b). A couple with no symptoms of sickle cell anaemia want to have a child. Both individuals have close relatives with the disease.

(i) Explain how a genetic counsellor can use knowledge of family history to provide genetic advice to the couple.

---

[2]

(ii) Suggest **two** ethical concerns that may be associated with the genetic testing of embryos for sickle cell anaemia.

1

2

---

[2]

11. Freeman was working on developing drought-resistant varieties of alfalfa using selective breeding, but this has proved difficult.
Drought resistance depends on the ability to withstand several abiotic factors, such as high temperatures and high light intensity.

(i) Use your knowledge of inheritance to suggest why it is difficult to study the genetic basis of drought resistance.

[2]

(ii) Alleles of the *miRNA* 156 gene regulate a group of transcription factors in alfalfa. These transcription factors activate or inhibit promoters that control genes related to drought resistance.

Explain how the *miRNA* 156 gene could be used to investigate the genetic basis of drought resistance.

[2]

(iii) Scientists have made a plasmid that produces more of the *miRNA* 156 gene product than normal and want to use this to develop a drought-resistant alfalfa plant.
Explain how they could incorporate the plasmid into alfalfa cells.

[2]

12(a). A Robertsonian translocation is a type of chromosomal translocation in which the long arms of two chromosomes fuse together.

Fig. 31.1 shows this event occurring between chromosomes 14 and 21.

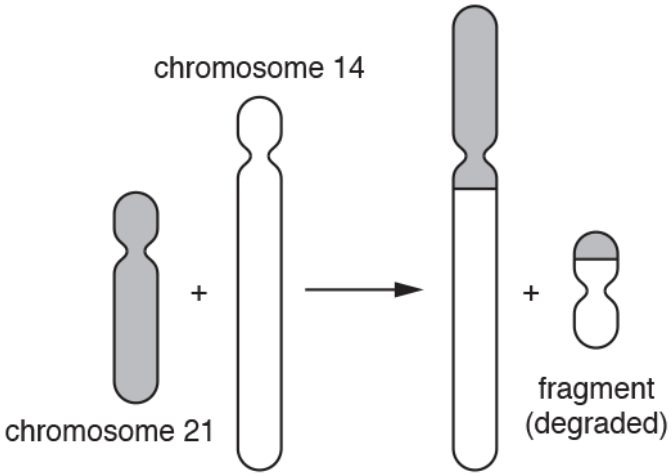


Fig. 31.1

An individual who inherits the translocated chromosome in Fig. 31.1 will either have Down's syndrome or be a carrier of the disorder.

A couple have a child. The mother is a carrier and the father is genetically normal. The genetic material with respect to chromosomes 14 and 21 in the somatic cells of the parents are shown in Fig. 31.2.

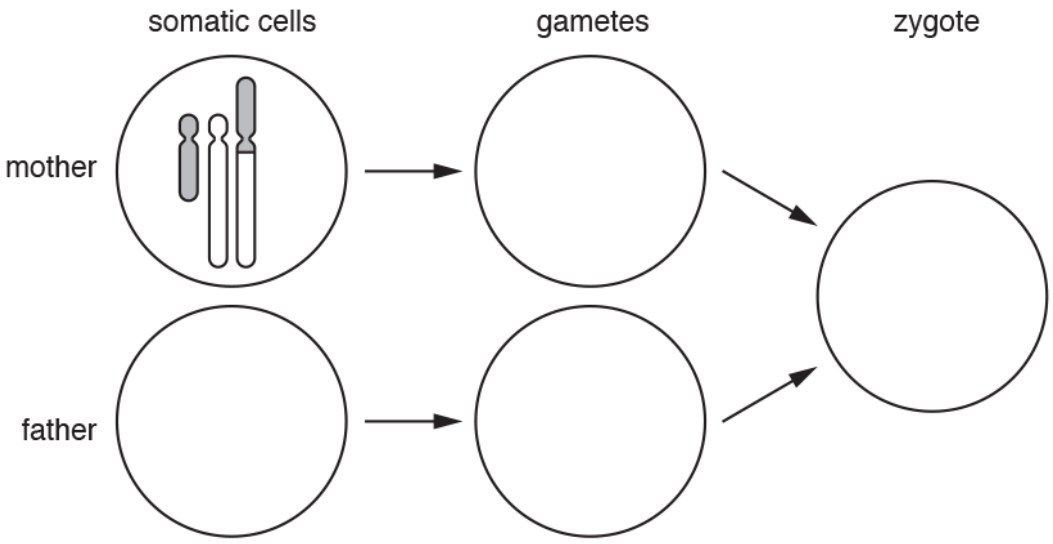


Fig. 31.2

(i) With reference to Fig. 31.2, suggest why the mother does **not** have Down's syndrome.

-- **[1]**

(ii) The child is born with Down's syndrome.

Complete the diagram in Fig. 31.2 to show the genetic material with respect to chromosomes 14 and 21 in:

- the somatic cell of the father
- the gametes of the mother and father
- the zygote of the child.

[Answer on Fig. 31.2] **[4]**

(b). Down's syndrome is more commonly caused by a genetic event that is distinct from that shown in Fig. 31.1.

State the name of this event **and** outline how it arises.

-- **[3]**

(c). A sample of cells can be collected from a fetus to test for genetic disorders such as cystic fibrosis.

(i) Chorionic villus sampling (CVS) and amniocentesis are two methods of obtaining fetal cells.

State the **source** of fetal cells that are obtained through these methods.

CVS ----------

Amniocentesis ----------

[1]

(ii) The sample of fetal cells can be used to produce a karyotype for genetic analysis.

Explain why karyotyping can **not** be used to detect cystic fibrosis.

---

[1]

END OF QUESTION PAPER

<table>
<tr><th colspan="3">Question</th><th>Answer/Indicative content</th><th>Marks</th><th>Guidance</th></tr>
<tr><td>1</td><td></td><td></td><td>Any 2 from:
(A and B are) codominant
idea that both genes are transcribed
idea that both antigen proteins are produced</td><td>2</td><td></td></tr>
<tr><td></td><td></td><td></td><td>Total</td><td>2</td><td></td></tr>
<tr><td>2</td><td></td><td>i</td><td>expected results correctly calculated (1)

<table><tr><th>F_1 phenotype</th><th>Observed results</th><th>Expected results</th></tr><tr><td>Smooth</td><td>547</td><td>549</td></tr><tr><td>Wrinkled</td><td>185</td><td>183</td></tr><tr><td>Total</td><td>732</td><td>732</td></tr></table>
Chi squared value correctly calculated (1)</td><td>2</td><td>$\chi^2 = \frac{(547-549)^2}{549} + \frac{(185-183)^2}{183}$

= 0.007 + 0.022

χ^2 = 0.029

ALLOW one mark for correct working if Chi squared value is incorrect
$\chi^2 = \frac{(547-549)^2}{549} + \frac{(185-183)^2}{183}$

ALLOW one mark for incorrect expected results used correctly in the equation to give a Chi squared value</td></tr>
<tr><td></td><td></td><td>ii</td><td>no significant difference between the observed and expected
results / do not reject null hypothesis (1)
test / chi squared value is smaller than the critical value at p = 0.05 (1)</td><td>2</td><td>To one degree of freedom
ALLOW accept null hypothesis
ALLOW ecf from (i) at correct degree of freedom</td></tr>
<tr><td></td><td></td><td>iii</td><td>heterozygote / heterozygous (1)

presence of wrinkled seeds in F1 (1)</td><td>2</td><td>ALLOW contains both dominant and recessive alleles
IGNORE any letters used in place of alleles</td></tr>
<tr><td></td><td></td><td></td><td>Total</td><td>6</td><td></td></tr>
<tr><td>3</td><td>a</td><td></td><td>1. polypeptide / protein / glycoprotein;
2. antigen;
3. loci;
4. cross(ing) over;
5. meiosis;
6. codominant;</td><td>6</td><td>2. ACCEPT receptor
3. ACCEPT ‘locus or locuses’
4. CREDIT ‘chiasma / chiasmata

Examiner's Comments

This question proved to be very accessible to candidates. This was done well although some candidates failed to carry the information forward to the next part of the question.</td></tr>
</table>

Question			Answer/Indicative content	Marks	Guidance
	b		1. ref to MHC genes and ABO genes being, unlinked / on separate chromosomes / on chromosomes 6 and 9; 2. ref to independent assortment (of chromosomes 6 and 9) in meiosis; 3. *idea that* gametes(s) contain same copy of chromosome 6 / different copies of chromosome 9; 4. children could be blood group O or A; 5 ref to 25% probability that a child could have, blood group O OR (only) 75% probability of, blood group A OR children could be ii / I^oI^o , I^AI^A, or I^A i 6. ref to 25% chance of having same haplotype;	3 max	5. CREDIT this mark point on a genetic diagram Examiner's Comments Several candidates misread the question and assumed that it was similarities and differences between the parents and children that were being referred to. Since they had just completed a genetic diagram showing that this does not happen this was disappointing. Many candidates were able to explain how the children could have different blood groups although the term 'heterozygous' triggered some candidates to respond in terms of the AB blood group. The fact that the MHC alleles and the blood group alleles were on different chromosomes was spotted by several candidates but relatively few went on to discuss independent assortment and some referred to crossing over, indicating a basic misconception about which chromosomes are involved in crossover events.
			Total	9	

Question			Answer/Indicative content	Marks	Guidance
4	a	i	e;	1	CREDIT X^e ACCEPT a single, lower case alternative letter IGNORE 'c' unless it is clearly 'lower case' IGNORE I^e
		ii	X^eY and X^eX^e	1	ecf for the allele symbol in **(i)**
		iii	X^EX^e	1	ecf for the allele symbol in **(i)** Examiner's Comments While many were aware that 'e' would be the allele symbol for the condition, some put a genotype (ee) in (i) and only the better candidates went on to give the correct genotypes for (ii) and (iii).
	b	i	1. nucleus / nucleolus; 2. ribosomes;	1	ANSWERS MUST BE IN THIS ORDER ACCEPT RER / rough endoplasmic reticulum
		ii	*idea that* mutation (in DNA) leads to a, stop / termination, codon OR *idea that* DNA sequence is deleted OR idea that mutation leads to more RNA being spliced out; *idea that* translation is terminated by stop codon; no (more) amino acids are added (after the stop codon) / fewer amino acids (in truncated protein);	2 max	IGNORE ref to a base deletion in context of shorter DNA Examiner's Comments Part (i) was synoptic and answered well but in (ii) too many answers were given in terms of changing the sequence of amino acids rather than producing a protein with fewer amino acids.
	c	i	reverse transcriptase;	1	
		ii	somatic (gene therapy);	1	CREDIT augmentation (gene therapy)

Question			Answer/Indicative content	Marks	Guidance
		iii	*Idea that* disease involves a defect in a single gene; no other cause; *idea that* disease is easily identified in sufferers; gene / gene product, is known; gene can be delivered reliably to affected cells;	3	Mp2 ACCEPT idea that disease will be cured / vision will be restored. Mp3 CREDIT reverse argument 'easy to see if it has worked' ACCEPT 'easy to insert the gene' Examiner's Comments This was intended to be accessible so it was disappointing how many candidates did not 'spot' reverse transcriptase or the fact that this would be somatic gene therapy. In part (iii) many candidates did not pick up on the question rubric and answered in terms of germ line therapy or ethics. It would be a worthwhile discussion for Centres to have with their candidates as to what makes some genetic diseases targets for treatment using gene therapy while others are not.
			Total	12	

Question			Answer/Indicative content	Marks	Guidance
5			Huntington's (disease); PKU / phenylketonuria; haemophilia; nail patella syndrome; muscular dystrophy / DMD; thalassaemia; AVP;;	2	**Mark the first answer on each prompt line.** If the answer is correct and an additional answer is given that is incorrect or contradicts the correct answer = **0 marks** **IGNORE** 'colour blindness' **DO NOT CREDIT** Turner's syndrome / Down's syndrome / Klinefelters syndrome Examiner's Comments This was done well although some candidates did not appreciate the significance of the term 'allele' in the question and gave answers which included Down's, Turner's or Klinefelter's. Very astute candidates gave familial glycosuria as an answer and this was credited. Hunter's syndrome was given as a response. This is a genetic disease and was credited although it was possibly an error on the part of the candidate. The question asked for two diseases to be stated rather than named so PKU was acceptable as were phonetic spellings although they needed to be approaching 'phenylketonuria'.
			Total	**2**	

Question			Answer/Indicative content	Marks	Guidance
6	a		*karyotypes* **K1** 2.1a has 46 chromosomes / DS karyotype has 47; **K2** 2.1a has 2 copies of chromosome 21 / DS karyotype has 3; **K3** 2.1a Chromosome 14 copies are different sizes / DS chromosome 14 same size; 1 (translocation) occurs during meiosis; 2 chromosomes / chromatids, break and rejoin; 3 *idea that* a piece of chromosome 21 attaches (to chromosome 14); 4 both copies of chromosome 14 and 21 segregating independently; 5 *idea that* one gamete has (both) chromosome 21 an additional piece of chromosome 21; 6 *idea that* (this) gamete, fertilised by / fertilises, a normal gamete;	6	**ACCEPT** a stage in meiosis e.g. prophase 1 **CREDIT** mps 2 to 6 if shown on annotated diagrams. **CREDIT** sperm or oocyte for gamete in mps 5 and 6 **CREDIT** description of a normal gamete e.g. one copy of 21 **ACCEPT** mp6 in context of Trisomy 21

Question			Answer/Indicative content	Marks	Guidance
			QWC;	1	LOOK FOR ONE mark from K1 - K3: AND mps 5 and 6 Examiner's Comments This was split between AO1 and AO2 criteria. This question was split into two parts. The first part required candidates to 'spot' differences between the karyotype shown and that of DS due to Trisomy 21. For many candidates, this was the only mark they achieved. Some weaker candidates confused DS with Turner's or Klinefelter's syndrome. Many candidates could successfully describe what happens in a translocation although some were describing the process but calling it a non-disjunction. However, very few candidates realised that, following translocation, the 'extended' chromosome 14 could segregate with the 'normal' 21 and that it was the fertilisation of a gamete with this combination of chromosomes by a 'normal' gamete that would lead to this karyotype. Candidates are clearly familiar with this inherited form of DS but need far more guidance as to how it is actually inherited.

Question			Answer/Indicative content	Marks	Guidance
	b	i	*idea that* a reason given for information being sensitive;	1	Look for ideas such as: 'women might not want people to know because they feel guilty OR people don't want to be discriminated against or judged OR to comply with legislation e.g. on safeguarding DO NOT CREDIT ideas about 'privacy' without some further qualification. Examiner's Comments This question addressed AO3 criteria. Weaker candidates referred to people 'wanting' to keep details private which was not enough to gain credit. Some further explanation was required and some very perceptive and thoughtful answers were seen.

Question			Answer/Indicative content	Marks	Guidance
		ii	improvements in (antenatal) screening techniques / AW; increase in, population / birth rates / pregnancies; *idea that* more women now screened; more older women having children / AW; *idea that* increasing age is risk factor (for DS);	2	CREDIT example such as introduction of nuchal fold screening or serum testing or 'triple' test in antenatal care IGNORE reference to improvements in karyotyping ACCEPT improvements in medical technology ACCEPT idea that younger women are now being screened. Examiner's Comments In **(ii)** explanations were required and the commonest answers referred to either increased maternal age or increase in the screening. Good candidates also spotted that the data was not a rate and correctly suggested increasing birth rates. Weaker candidates described the data without necessarily explaining.
		iii	*idea of* fetal viability / AW;	1	CREDIT statements which refer to increase chance of a fetus with DS miscarrying naturally Examiner's Comments Part **(iii)** was a stretch and challenge question and many candidates correctly deduced that the viability of fetus with DS would have to be taken into account. Again, some candidates expressed their ideas with great sensitivity.
			Total	11	

Question			Answer/Indicative content	Marks	Guidance
7		i	7 (µm);;	2	Correct answer = 2 marks If answer is incorrect, award one mark for correct working -(2 or 20 or 20 000) ÷ 2 800 OR (1.9 or 19 or 19 000) ÷ 2 800 Award one mark for an answer which has not been rounded 7.142……. OR 6.785…. Examiner's Comments The calculation in (i) was done well by many candidates although many did not notice that the answer was required in µm or used incorrect factors of 10 in their calculations. Part (i) was synoptic with candidates needing to recall the role of platelets and suggest how this could cause red blood cell fragmentation.
		ii	*idea that* narrow lumen / roughened endothelium / blood clot, causes (red blood cells to fragment);	1	ACCEPT description of blood clot e.g formation of fibrin mesh causing fragmentation of red blood cells Examiner's Comments In (ii) a surprising number of candidates opted for haemophilia - possibly triggered by the platelets and blood clotting ideas in the previous part of the question.
		iii	sickle cell (anaemia) / AVP;	1	Examiner's Comments (iii) tested entirely AO1 objectives.
			Total	4	

Question			Answer/Indicative content	Marks	Guidance
8		i	interrupts triplet code / causes frame-shift / production of premature stop codon ✓ (DNA) triplets code for different amino acids ✓ primary structure of protein changed ✓ protein cannot interact with other, proteins / named component ✓	max 2	
		ii	gene (encoding Factor VIII), on X chromosome / X-linked / sex-linked ✓ males lack second X chromosome ✓	2	ORA ORA
		iii	male gametes: X^h, Y female gametes: X^H, X^h X^HX^h | X^HY X^hX^h | X^hY ✓✓ probability = 0.5 ✓	3	ALLOW H/h not superscripted on X ALLOW 50% or ½
			Total	7	

Question			Answer/Indicative content	Marks	Guidance
9		i	25 (%);	1	IGNORE working Examiner's Comments The majority of candidates were able to calculate the theoretical percentage of a heterozygous individual being produced in a cross between two heterozygous individuals.

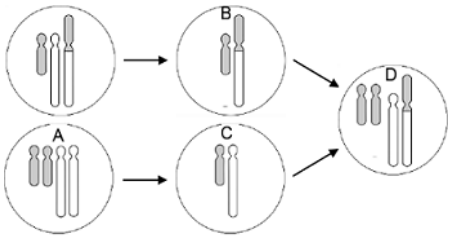
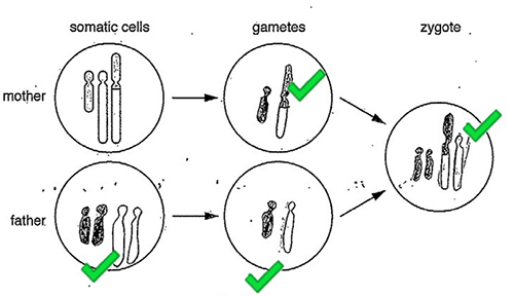
Question			Answer/Indicative content	Marks	Guidance
		ii	1. (island edges / cacti) subject to, sea/salt, spray; 2. qq (genotype) confers ability to obtain water from salt spray; 3. (gives) selective advantage; 4. (individuals with qq genotype) survive / reproduce; 5. allele / q, frequency increases; 6. directional selection; 7. geographic, isolation / barrier; 8. (means) no new alleles coming in;	4 max	ACCEPT ORA for mp 2 – 5 IGNORE mist / sea water for mp1 and 2 ACCEPT homozygous recessive / 'they' for qq genotype 2. ACCEPT qq gets water supply from salt spray 2. ACCEPT qq genotype confers tolerance to salt (spray) 3. ACCEPT description e.g. 'they are (at an advantage and are) selected for' 5. DO NOT CREDIT gene frequency increases 5. IGNORE 'qq frequency increases' 6. IGNORE natural selection Examiner's Comments Good responses were able to comprehensibly explain how individuals homozygous for the q allele would come to dominate the gene pool of the *Brachycereus* species of cactus in the Galapagos Islands. It was essential to refer to the local conditions where the cactus is subjected to salty sea spray. Candidates realising the prevailing conditions, correctly referred to the qq individuals being able to obtain water from the salty spray, giving them a selective advantage and allowing them to survive. A few acceptable alternative arguments were also seen for the other cacti with the Q allele being unable to thrive. A number of responses failed to go as far as explaining that this selective advantage would result in the frequency the q allele rapidly increasing. Rarely was it mentioned that this was directional selection or that the cactus was geographically isolated with no new alleles coming into the population.
			Total	5	

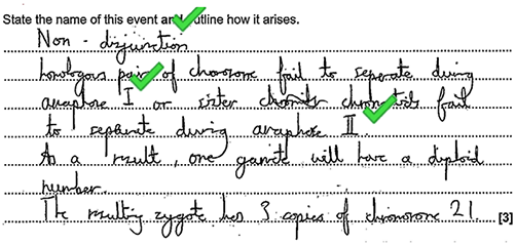
Question			Answer/Indicative content	Marks	Guidance
10	a		substitution ✓	1	ALLOW ref to single base replacement. IGNORE point mutation Examiner's Comments (a) addressed AO1 criteria with the remaining parts of the question addressing AO2 and some AO3 in (b)(iii). (a) was generally done well.
	b	i	(construct) pedigree / genetic tree (diagram) ✓ to calculate, probability / chance / likelihood of disease inheritance ✓	2	IGNORE ref to genetic testing Examiner's Comments There were two common incorrect responses in (b)(i). Many candidates suggested that the role of the genetic counsellor was to work out the genotype of the parents rather than predict the probability of their child being born with a genetic disease. Secondly, there were some inaccurate descriptions of how they might do this referring to a genetic cross, Punnet square or family tree rather than the more scientific term of pedigree analysis.
		ii	any 2 from: may lead to abortion / decision as to whether child should be born ✓ damage to embryo and / or mother ✓ false positive / false negative, results ✓ idea that embryo cannot give consent ✓	max 2	 ALLOW increased risk of miscarriage DO NOT ALLOW inaccuracy unqualified IGNORE ref to playing God Examiner's Comments Most candidates scored at least one mark for part (b)(ii) and did restrict their answers to ethical considerations. However, the question asked about testing for sickle cell anaemia and therefore more generalised responses suggesting that this might lead to designer babies were not credited.
			Total	5	

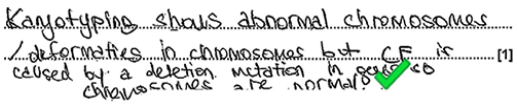
Question			Answer/Indicative content	Marks	Guidance
11		i	(drought resistance) involves many, genes / loci ✓ Involves (possible), multiple / variety, alleles (at each locus) ✓	2	ALLOW (drought resistance) is polygenic IGNORE different alleles Examiner's Comments Some candidates were able to appreciate the polygenic aspect of drought resistance and successfully linked the information in the stem of the question with genes and inheritance. Many candidates did not recognise the relevance of 'inheritance' and 'genetic basis' in the stem of the question and discussed their answers in terms of phenotype only e.g. not many plants survive, so there are very few that can be studied.
		ii	(gene/ transcription factor) can be used to identify, genes / alleles, involved / activated, in drought resistance ✓ promoters allow, gene expression / transcription ✓ remove / knockout, (miRNA 156) gene and observe, phenotype / drought resistance ✓	2	ALLOW RNA Polymerase binds to promoters Examiner's Comments Candidates struggled to understand the relevance of the miRNA gene and confused this with RNA. However, many candidates did discuss knocking out miRNA to observe the phenotype and so exhibited a good understanding of knockout technology.

Question			Answer/Indicative content	Marks	Guidance
		iii	place plasmid in Agrobacterium / use gene gun ✓ screen for, transgenic/ recombinant, cells ✓ details of selectable marker ✓	2 max	ALLOW use *Agrobacterium* as vector for 'place plasmid in *Agrobacterium*'. ALLOW a description of a gene gun IGNORE ref to viruses / liposomes IGNORE ref to antibiotics <u>Examiner's Comments</u> Few candidates scored full marks. Those that scored one mark usually referred to a gene gun for insertion of the plasmid. Most candidates did not appreciate that the alfalfa would be insertion into plant cells and referred to vectors relevant to animal cells e.g. viruses. There were a lot of descriptions using restriction enzymes and ligases to obtain the recombinant plasmid, not realising the focus of the question was on the incorporation of the plasmid.
			Total	6	

Question			Answer/Indicative content	Marks	Guidance
12	a	i	does not have trisomy 21 ✓	1	AW e.g. 'no excess of chromosome 21' ALLOW only has two of chromosome 21 Examiner's Comments This question required candidates to know that Down's syndrome is a disorder caused by extra genetic material relating to chromosome 21. This can be when there are three distinct copies of chromosome 21(trisomy 21) or some extra material relating to chromosome 21. In this case the mother does not have any extra genetic material and therefore does not have Down's syndrome.

Question			Answer/Indicative content	Marks	Guidance
		ii	 A ✓ B ✓ C ✓ D ✓	4	ALLOW incorrect proportions of translocated chromosome ALLOW diagrams that do not show centromeric regions DO NOT ALLOW drawings without shading ECF (from A) ECF (from B and C) <u>Examiner's Comments</u> Very few candidates were able to complete all four of the diagrams correctly. Many candidates were not able to identify which chromosomes would be present in the gamete of the mother but were credited with subsequent marks if they understood that the zygote would result from a combination of the two gametes. Candidates need to be careful when completing diagrams such as these to make sure that they include the relevant shading or annotation to distinguish the relevant chromosomes. **Exemplar 1**  In this response the candidate completed the diagrams correctly. The shape and shading of the chromosomes was clear and unambiguous.

Question			Answer/Indicative content	Marks	Guidance
	b		(meiotic) non-disjunction ✓ any 2 from: failure of homologous chromosomes to separate during meiosis I/anaphase I ✓ failure of sister chromatids to separate during meiosis II/anaphase II ✓ gamete has extra copy of chromosome (21) ✓	max 3	ALLOW for one mark - failure of homologous chromosomes or sister chromatids to separate during meiosis Examiner's Comments Few candidates were able to recall non-disjunction, but the question also required a precise description of what might happen during meiosis to result in the formation of a zygote with three chromosomes 21s. Exemplar 2  State the name of this event and outline how it arises. Non - disjunction homologous pair of chromosomes fail to separate during anaphase I or sister chromatids chromatids fail to separate during anaphase II As a result, one gamete will have a diploid number The resulting zygote has 3 copies of chromosome 21 [3] This response recalls the name and gives a succinct and clear description of events of meiosis. There was an implication that all the chromosomes failed to separate making the gamete diploid and therefore this response would not have been credited with the idea of the gamete having an extra chromosome 21. However, it gained the maximum of three marking points in the first four lines.
	c	i	CVS: placenta amniocentesis: amniotic fluid ✓	1	IGNORE chorionic villus Both required for 1 mark

Question			Answer/Indicative content	Marks	Guidance
		ii	1 from: (karyotype) cannot detect gene/allele (mutations) ✓ (karyotype) can only detect changes in chromosome size/shape ✓	1	AW e.g. abnormal base sequence Examiner's Comments Most candidates were able to identify the source of fetal cells. In order to explain why karyotyping cannot be used to test for CF, candidates had to have some knowledge of the cause of CF. Exemplar 3  Karyotyping shows abnormal chromosomes / deformities in chromosomes but CF is caused by a deletion mutation in gene so chromosomes are normal? [1] This response describes the cause of CF. Any idea of CF being a gene mutation would be sufficient. The response also explains why it would not be picked up on a karyotype.
			Total	10	