Q1.

4 Fig. 4.1 is a diagram drawn from a photomicrograph of an animal cell undergoing meiosis.

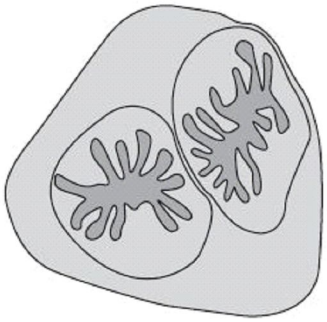


Fig. 4.1

(a) Identify the stage of meiosis shown in Fig. 4.1.

.....[2]

(b) Describe the main events that will occur to complete meiosis from this stage.

.....

.....

.....

.....

.....[4]

(c) Describe **two** ways in which meiosis leads to variation.

.....

.....

.....

.....

.....

[4]

[Total : 10]

Q2.

5 (a) Distinguish between phenotype and genotype.

.....

.....

.....

.....[2]

(b) Describe how artificial selection differs from natural selection.

.....

.....

.....

.....[3]

(c) Define the terms

(i) *gene;*

.....

.....

.....[2]

(ii) *allele.*

.....

.....

.....

.....[3]

[Total : 10]

Q3.

5 Coat colour in cats is determined by a sex-linked gene with two alleles, black and orange. When black cats are mated with orange cats, the female offspring are always tortoiseshell, their coats show black and orange patches of various sizes, while the male offspring have the same coat colour as their mothers.

(a) Using the symbols X^B for black and X^O for orange, draw genetic diagrams to account for both these crosses.

black female X orange male	orange female X black male	

(b) List the genotypes and their phenotypes of the offspring that may result from mating a tortoiseshell female with a black male.

.....

.....

.....

.....

.....[4]

(c) Suggest an explanation for the tortoiseshell coat in terms of the activity of the X chromosomes.

.....

.....

.....[1]

[Total: 9]

Q4.

2 Fig. 2.1 is a diagram of pair of homologous chromosomes during meiosis.

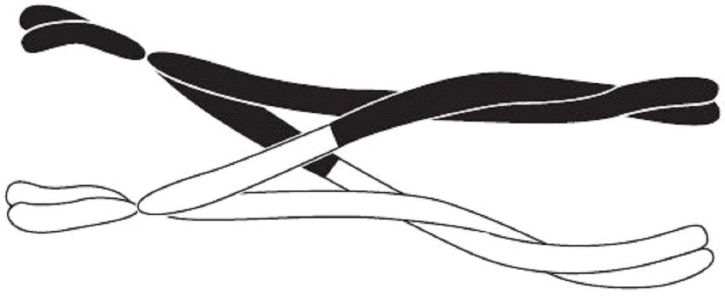


Fig. 2.1

(a) State what stage of meiosis is shown.

.....[1]

(b) Describe what has occurred between the two homologous chromosomes.

.....

.....

.....

.....

.....[3]

(c) Explain how this can lead to variation.

.....

.....

.....[2]

(d) Describe **two** other sources of variation that are possible as a result of meiosis.

.....

.....

.....

.....

.....

.....[4]

[Total: 10]

Q5.

5 (a) The summer squash plant produces fruit that are either white or yellow in colour and are either shaped like a disc or a sphere. The dominant phenotypes are white and disc-shaped fruit. Using the symbols **A** for white and **a** for yellow and **B** for disc and **b** for sphere, draw a genetic diagram to show what proportion of offspring will have yellow and sphere-shaped fruit if a white and disc-shaped fruit plant, heterozygous for both genes, is self-fertilised.

Sickle cell anaemia is a blood disease that is frequently fatal when homozygous. It is caused by an autosomal recessive allele. Heterozygotes have sickle cell trait and appear normal.

Malaria is a potentially fatal infectious disease of the blood caused by the protoctist, *Plasmodium*. In parts of the world where malaria is endemic the frequency of the sickle cell allele is high.

(b) Explain the possible health consequences, in such areas, for a person who is homozygous dominant and for a person who is homozygous recessive for the sickle cell allele.

(i) homozygous dominant for the sickle cell allele

.....

.....[1]

(ii) homozygous recessive for the sickle cell allele.

.....

.....[1]

(c) Explain why heterozygotes have a strong selective advantage in areas where malaria occurs.

.....

.....

.....

.....

.....

.....[3]

[Total: 11]

Q6.

7 **(a)** Sometimes a gene has more than two alleles, termed *multiple alleles*.
The ABO blood group system in humans is controlled by a gene with three alleles, I^A, I^B and I^o. Alleles I^A and I^B are codominant and I^o is recessive to both.

The blood group **AB** is the result of codominance.

Explain what is meant by *codominance*.

.....

.....

.....

.....

.....[3]

(b) In humans, a gene that codes for the production of a protein, called factor VIII, is located on the X chromosome. The dominant allele for this gene produces factor VIII, but the recessive allele does not produce factor VIII.

A person who is unable to make factor VIII has haemophilia in which the blood fails to clot properly.

Explain why a man with haemophilia cannot pass haemophilia to his son but may pass haemophilia to his grandson.

.....

.....

.....

.....

.....

.....[3]

(c) A gene for feather colour in chickens is carried on an autosome. This gene has two alleles, black (C^B) and splashed-white (C^W). When a male chicken with black feathers is mated with a female chicken with splashed-white feathers, all the offspring have blue feathers. This also occurs when a male chicken with splashed-white feathers is crossed with a female with black feathers.

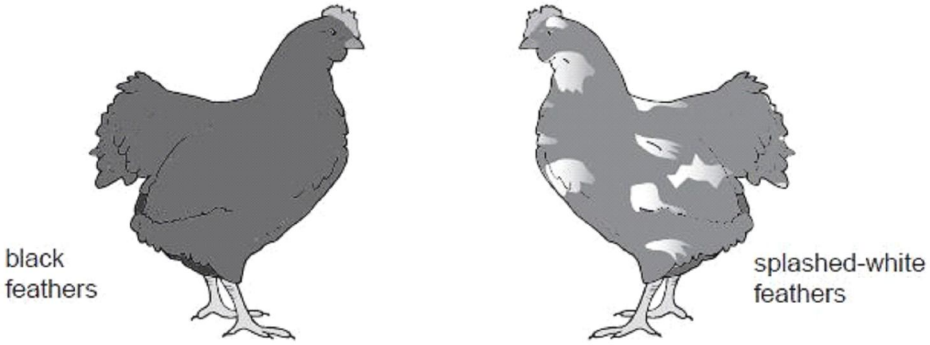


Fig. 7.1

Another gene may cause stripes on feathers (barred feathers). This gene is carried on the X chromosome. The allele for barred feathers (X^A) is dominant to the allele for non-barred feathers (X^a).

In chickens the male is homogametic and has two X chromosomes while the female is heterogametic and has one X chromosome and one Y chromosome.

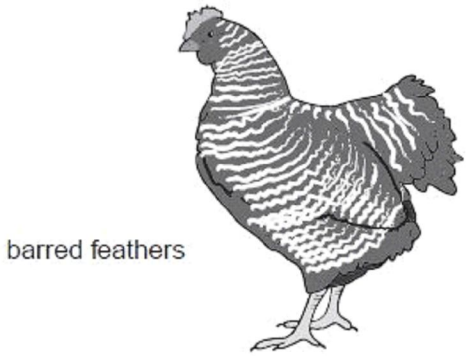


Fig. 7.2

(i) A male chicken with black, non-barred feathers was crossed with a female chicken with splashed-white, barred feathers. All the offspring had blue feathers, but the males were barred and the females were non-barred.

Using the symbols given above draw a genetic diagram to show this cross.

parents' phenotype	male, black, non-barred feathers.	female, splashed-white, barred feathers.
genotype		
gametes		
offspring genotypes		
phenotypes	male, blue, barred feathers.	female, blue, non-barred feathers.

[5]

(ii) Explain how a farmer could use a breeding programme to find out the genotype of a male chicken with blue, barred feathers.

.....

.....

.....

.....

.....

.....[3]

[Total: 14]

Q7.

6 Colour blindness is a condition characterised by the inability of the brain to perceive certain colours accurately.

- The most common form is termed red-green colour blindness (RGC).
- RGC results from a recessive allele.
- 0.6% of females worldwide have RGC.
- 8.0% of males worldwide have RGC.

Fig. 6.1 shows the occurrence of RGC in one family.

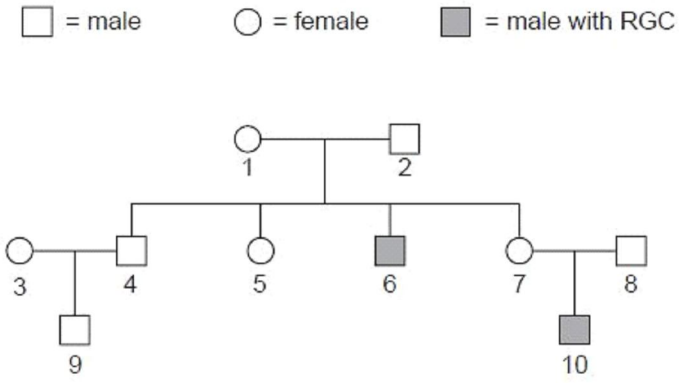


Fig. 6.1

(a) Explain the meaning of the terms *allele* and *recessive*.

allele .....

.....[1]

recessive .....

.....[1]

(b) Explain why females are less likely than males to have RGC.

.....

.....

.....[2]

(c) With reference to Fig. 6.1, and using the symbols **R** for the dominant allele and **r** for the recessive allele, state the genotypes of the individuals **1**, **4**, **6** and **7**.

1 .....

4 .....

6 .....

7 ..... [4]

[Total: 8]

Q8.

7 **(a)** The inheritance of coat colour in horses is complex but all horses have one of two base colours, red (chestnut) or black. The base colour is controlled in a simple monohybrid way.

- When chestnut stallions and mares are mated the foals are always chestnut.
- When black stallions are mated with black mares, either black or chestnut foals may be produced.

Draw a genetic diagram to show how two parents with black coat colour can produce a chestnut foal **and** the probability of such an event occurring.
Choose a letter symbol to represent coat colour.

For Examin Use

[4]

(b) Five other genes can modify the base coat colour.

One of these genes is the **C** gene. There are two alleles of this gene, **C** and $\mathbf{C}^{CR}$.

- **C** does not affect the base coat colour.
- $\mathbf{C}^{CR}$ may modify the base coat colour.
- If a chestnut horse has at least one $\mathbf{C}^{CR}$ allele its phenotype will be palomino, which is a light cream colour.
- If a black horse has at least one $\mathbf{C}^{CR}$ allele its effect will not be noticeable in the phenotype.

Complete the genetic diagram below.

parental genotype	**aaCC^{CR}**		**AaCC**		
parental phenotype	.....		.....		
gametes	.....	.....	.....	.....	
offspring genotypes	.....	.....	.....	.....	
offspring phenotypes	.....	.....	.....	.....	[4]

[Total: 8]

Q9.

6 **(a)** Explain what is meant by the term *gene mutation*.

.....

.....

.....

..... [2]

(b) Rickets is a childhood disorder involving the softening and weakening of bones. It is usually caused by a lack of vitamin D, calcium ions or phosphate ions. A rare form of rickets that cannot be successfully treated with vitamin D therapy is caused by a mutant allele on the X chromosome.

Fig. 6.1 shows a pedigree chart for a family that has a history of this condition.

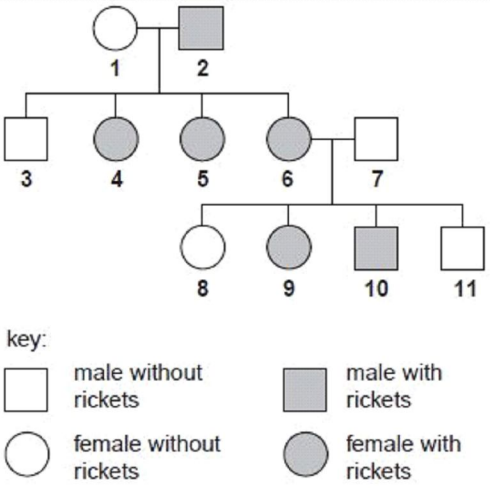


Fig. 6.1

Using the symbols

X^R for the mutant allele on the X chromosome
X^r for the non-mutant allele on the X chromosome

state the genotypes of the following individuals.

1 .....

3 .....

9 .....

10 ..... [4]

(c) The gene in which this mutation occurs codes for a protein found in the cells of the proximal convoluted tubule of the kidney. This protein is involved in phosphate ion transport across membranes.

Suggest why individuals with this mutant allele show symptoms of rickets.

.....

.....

.....

..... [2]

[Total: 8]

Q10.

6 In humans a rare, sex-linked, recessive allele results in a change in the shape of the iris in the eye. This condition is know as cleft iris (CI).

(a) Explain what is meant by the term *sex linkage*.

.....

.....

.....

..... [2]

(b) Using suitable symbols complete the genetic diagram below.

Key to symbols

recessive allele .....

dominant allele .....

parental phenotypes	male with CI	X	normal female
parental phenotypes	.....		.....
gametes	.....		.....

offspring genotypes .....

offspring phenotypes ..... [5]

(c) A woman who is heterozygous for CI becomes pregnant by a man with a normal iris.

State the probability that their child will have CI.

..... [1]

[Total: 8]

Q11.

6 Canavan disease is a non sex-linked inherited condition that causes progressive damage to neurones of the brain. Symptoms of the condition include a loss of motor skills and mental retardation. The symptoms appear in early infancy and many children with this condition die by the age of four years.

People with Canavan disease lack an enzyme called aspartoacylase which breaks down N-acetyl aspartate. The build up of N-acetyl aspartate can interfere with the formation of the myelin sheath, particularly in neurones of the brain.

(a) Enzymes such as aspartoacylase display specificity.

Outline what is meant by *specificity* of an enzyme.

.....

.....

.....

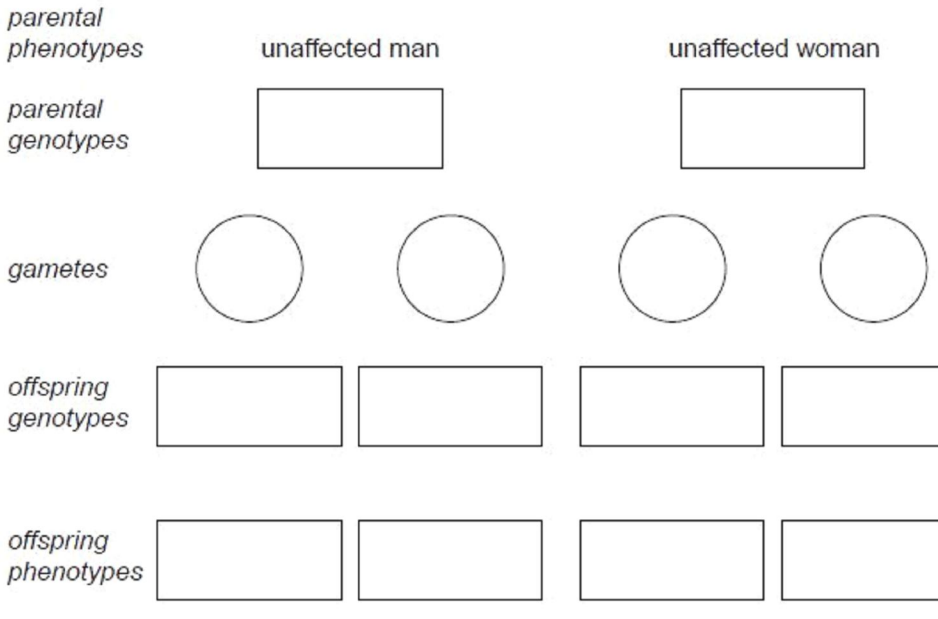
..... [2]

(b) Complete the genetic diagram below to show how an unaffected man and an unaffected woman could produce a child with Canavan disease.

key to symbols

.....

.....



[3]

(c) Explain the importance of the myelin sheath in the functioning of a neurone.

Exa

.....

.....

.....

.....

.....

.....

.....

..... [3]

[Total: 8]

Q12.

7 Meiosis is a type of nuclear division, which produces gametes for sexual reproduction.

(a) Fig. 7.1 shows diagrams of the stages of meiosis, **A** to **J**, but they are not in the correct order.

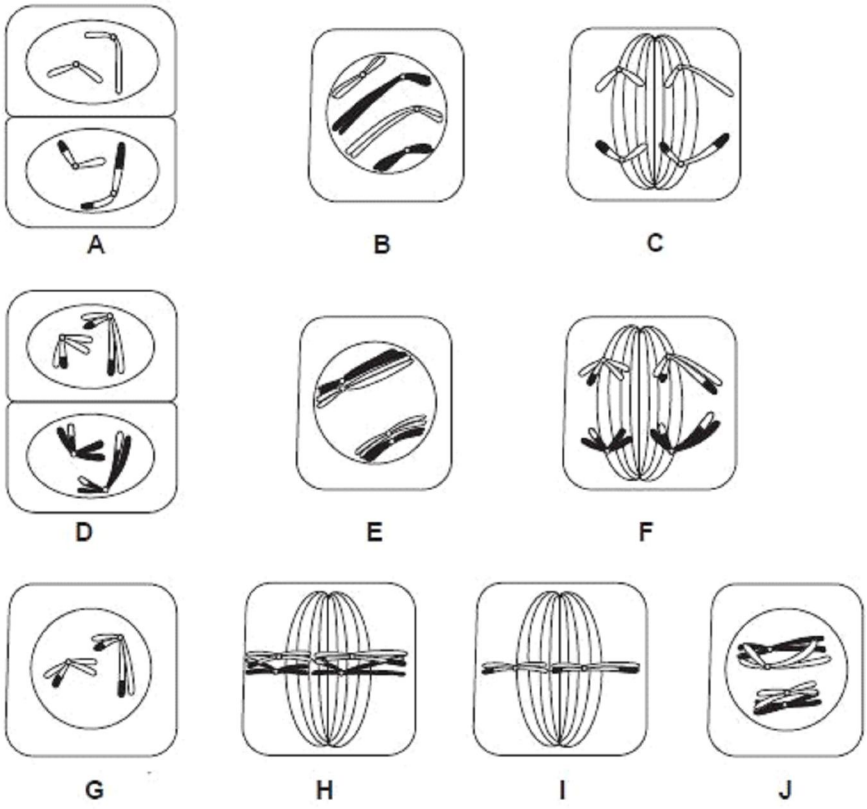


Fig. 7.1

Complete the table below by writing the stages of meiosis in the correct order.

Some of the stages have already been written in the table.

nuclear division	letter of stage
meiosis I	B
	D
meiosis II	

[4]

(b) Explain how meiosis can result in genetic variation amongst offspring.

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....[5]

[Total: 9]

Q13.

9 Huntington's Disease (HD) is a severe neurological disorder in which symptoms usually appear after the person has reached sexual maturity. Symptoms include memory loss and changes in personality and mood.

For Examiner Use

HD is caused by a gene mutation on chromosome 4 in which the triplet code CAG is repeated many times. The resulting allele is dominant.

(a) Explain what is meant by the terms gene mutation and triplet code.

gene mutation .....

.....

.....

.....

triplet code .....

.....

.....

.....[4]

(b) A couple wish to start a family. The man does not have HD but the woman does have the disease. The woman's father does not have the disease.

Complete the genetic diagram below to show the probability of the couple's first child having HD.

key
Huntington allele = T
normal allele = t

parental phenotypes	man without HD	woman with HD
parental genotypes	.....	.....
gametes	.....	.....
offspring genotypes	.....	
offspring phenotypes	.....	

probability of first child having HD ..... [3]

[Total: 7]

Q14.

7 The fruit fly, *Drosophila melanogaster*, has many phenotypic variations and has been used in experiments to demonstrate the principles of inheritance.

Fo
Exami
Us

(a) The majority of fruit flies have red eyes but there is a variant with white eyes.

Fig. 7.1 shows the red-eyed and white-eyed variants of the fruit fly.

Fig. 7.1

The gene for eye colour is located on the X chromosome.

Using suitable symbols, draw a genetic diagram to show the possible offspring of a cross between a heterozygous red-eyed female fruit fly with a white-eyed male fruit fly.

key to symbols:

.....

.....

parental phenotypes	red-eyed female	white-eyed male
parental genotypes		
gametes		
offspring genotypes		
offspring phenotypes		

[5]

(b) One of the genes controlling the clotting of blood in humans is also located on the X chromosome. A rare variation of the gene, a recessive allele for haemophilia, can lead to a condition where the blood fails to clot properly.

For Examin Use

(i) State why a man who has haemophilia is unable to pass the condition on to his son.

.....

.....[1]

(ii) Queen Victoria of Great Britain in the 19th century was a carrier of haemophilia, but did not have the condition.

State the term used to describe the genotype of a carrier.

.....[1]

(iii) Neither of Queen Victoria's parents carried the allele for haemophilia.

Suggest how Queen Victoria could have become a carrier.

.....

.....[1]

[Total: 8]

Q15.

7 **(a)** Explain what is meant by the term *heterozygous genotype*.

E

heterozygous .....

.....

genotype .....

.....[2]

(b) The budgerigar, *Melopsittacus undulatus*, is a small type of parrot that is native to Australia.

Fig. 7.1 shows a budgerigar.

Fig. 7.1

A budgerigar can have blue, green, yellow or white feathers.

Two genes, **A**/**a** and **D**/**d**, are involved in the inheritance of feather colour in budgerigars.

- A bird which has at least one dominant allele **A** but is homozygous for **d** has blue feathers.
- A bird which has at least one dominant allele **D** but is homozygous for **a** has yellow feathers.
- A bird with at least one dominant **A** allele **and** one dominant **D** allele has green feathers.
- A bird that is homozygous for **a** and **d** has white feathers.

(c) Two green-feathered budgerigars, heterozygous at both gene loci, were crossed.

Draw a genetic diagram of this cross to show the probability of producing offspring with yellow feathers.

[6]

Q16.

7 Coat colour in cats is determined by a sex-linked gene with two alleles coding for black and orange.

When black cats are mated with orange cats:

- the female offspring are always tortoiseshell (black and orange patches)
- the male offspring are always the same colour as their mother.

(a) Explain what is meant by a *sex-linked gene*.

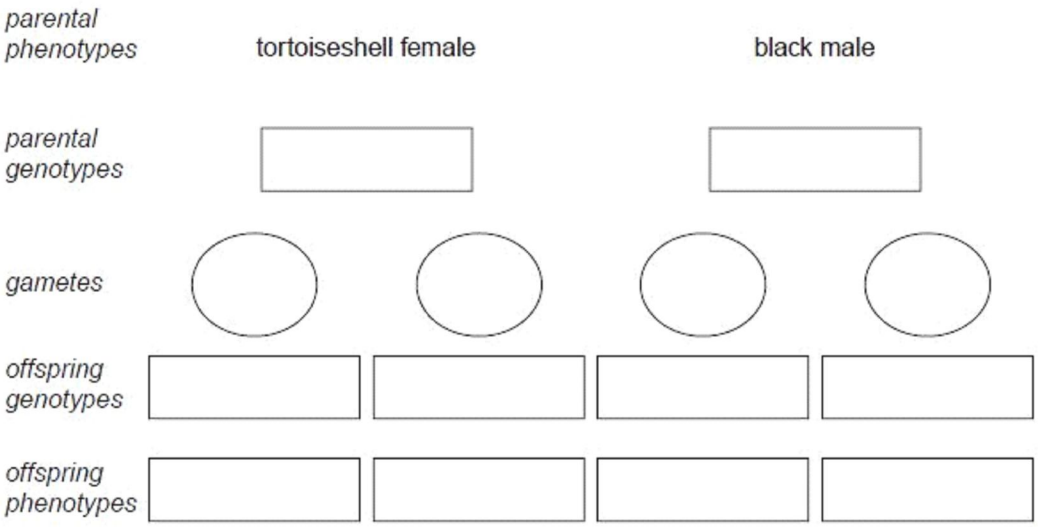
sex-linked .....

.....

gene .....

..... [2]

(b) Using the symbols **B** for the allele for black coat and **O** for the allele for orange coat, complete the genetic diagram below.



[4]

(c) Explain why a male cat cannot have a tortoiseshell coat.

.....

.....

.....

.....

..... [2]

[Total: 8]

Q17.

7 Resistance to the poison warfarin is now extremely common in rats. Warfarin inhibits an enzyme in the liver, vitamin K epoxide reductase, that is necessary for the recycling of vitamin K. This vitamin is involved in the production of substances required for blood clotting.

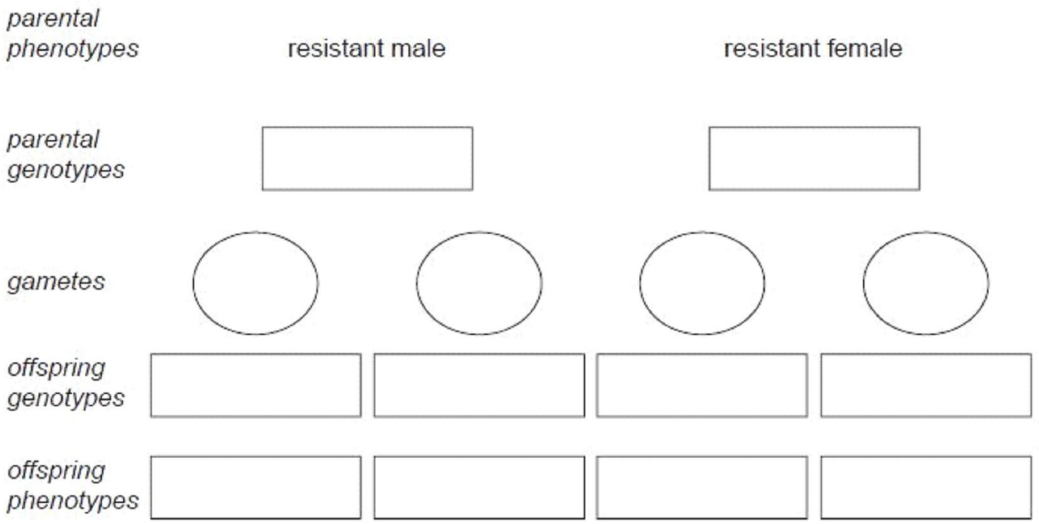
- Rats susceptible to warfarin die of internal bleeding.
- Rats that are homozygous for resistance to warfarin do not suffer from internal bleeding when their diet provides more than 70 μg of vitamin K per kg body mass per day.
- Heterozygous rats are resistant to warfarin when their diet provides about 10 μg of vitamin K per kg body mass per day.

(a) Using appropriate symbols, complete the genetic diagram to show how two resistant rats can produce warfarin-susceptible offspring.

key to symbols

.....

.....



[3]

(b) Rats that are homozygous for warfarin resistance have a low survival rate in the wild. Suggest why this is so.

.....

..... [1]

(c) Warfarin can be safely given to humans who are at risk of unwanted blood clots. The clotting time of the blood is measured regularly and the warfarin dose is varied accordingly.

Suggest, giving a reason, the type of inhibition warfarin has on the enzyme vitamin K epoxide reductase.

type of inhibition .....

reason .....

..... [2]

(d) The allele for warfarin resistance may have originated by a single base substitution and resulted in a modified vitamin K epoxide reductase.

Explain how a single base substitution may affect the phenotype of an organism.

.....

.....

.....

.....

.....

.....

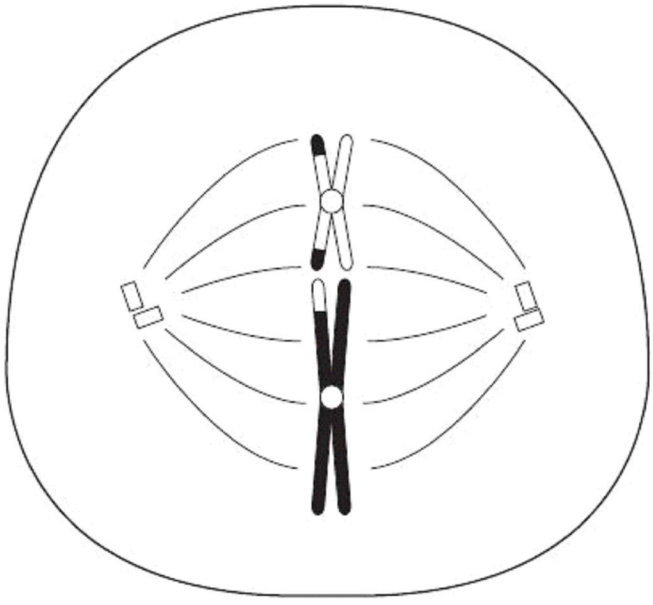
.....

..... [3]

[Total: 9]

Q18.

4 Fig. 4.1 shows the results of an animal cell that has undergone meiosis I.



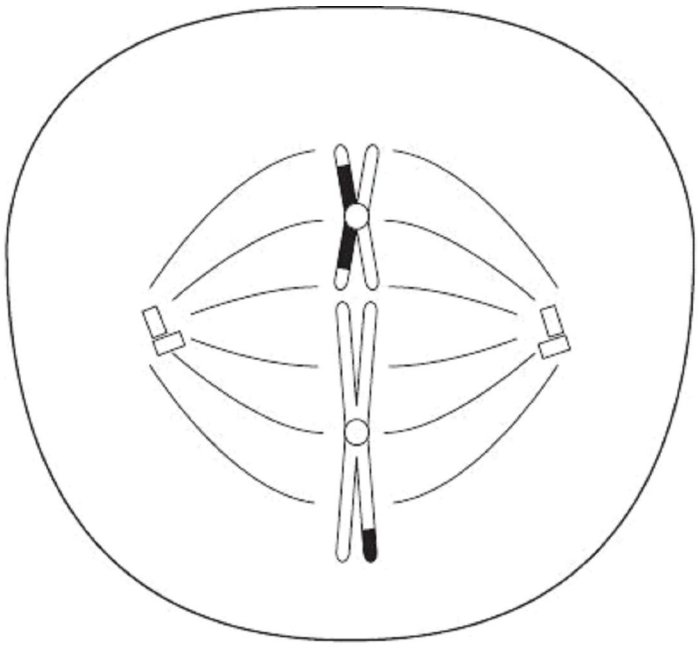


Fig. 4.1

(a) State which stage of meiosis II is shown.

.....[1]

(b) Describe the next stage of meiosis II.

.....

.....

.....

.....[3]

(c) Describe what has happened before the start of meiosis to

(i) the nuclear membrane;

.....

.....[1]

(ii) the centrioles.

.....

.....[2]

(d) Name and explain two ways in which meiosis can lead to variation.

1. .....

.....

.....

2. .....

.....

.....[4]

[Total : 11]

Q19.

5 Scallops, which are bivalve molluscs, are important commercially throughout the world. The marine bay scallop, *Agropecten irradians*, has three distinct shell colours, yellow, orange and black. The shell colour is controlled by a gene with three alleles, yellow, S^y, orange, S^o, and black, S^b.
Scallops are hermaphrodite and are able to fertilise themselves to produce offspring.
Single mature adult specimens of yellow, orange and black scallops were collected and kept in separate tanks of seawater until they produced young. The young were then scored for shell colour. The results were as follows.

yellow scallop – 25 yellow and 8 black
orange scallop – 31 orange and 9 black
black scallop – 27 black

(a) Explain the results from the orange and black scallops, using the symbols given.

.....

.....

.....

[6]

(b) Orange scallops are more valued for human consumption.

Describe how a marine biologist could produce a pure-breeding line of orange scallops for commercial exploitation using the offspring from the single orange scallop.

.....

.....

.....

.....[2]

[Total : 8]

Q20.

4 Resistance to the widely used poison warfarin is now extremely common in rats. Warfarin interacts with vitamin K to prevent its normal functions in the blood clotting mechanism. Normal rats fed on warfarin suffer a fatal haemorrhage. Resistant rats apparently do not use vitamin K in the same way and maintain normal blood clotting times, even when they have eaten large amounts of warfarin. Warfarin resistance in rats is determined by a single dominant allele. Animals carrying the allele for resistance need large quantities of vitamin K.

genotype	resistance to warfarin	quantities of vitamin K required
homozygous recessive	not resistant (susceptible)	normal
heterozygous	resistant	slightly higher
homozygous dominant	resistant	extremely large

When warfarin is used continually the percentage of resistant rats remains at about 50% of the total rat population.

(a) Using the symbols **R** for the allele that confers warfarin resistance and **r** for the allele that produces no resistance, draw a genetic diagram to explain how resistant rats can produce warfarin susceptible offspring.

[4]

(b) Suggest why homozygous dominant rats are unlikely to survive in the wild.

.....

.....[1]

(c) Describe how natural selection operates to maintain the proportion of resistant rats at about 50% of the total population.

.....

.....

.....

.....[3]

(d) Explain, with an example, how a mutation that results in the substitution of a single base may affect the phenotype of an organism.

.....

.....

.....

.....[3]

[Total : 11]

Q21.

4 Fig. 4.1 shows four generations of a family in which some members of the family suffer from sickle cell anaemia.

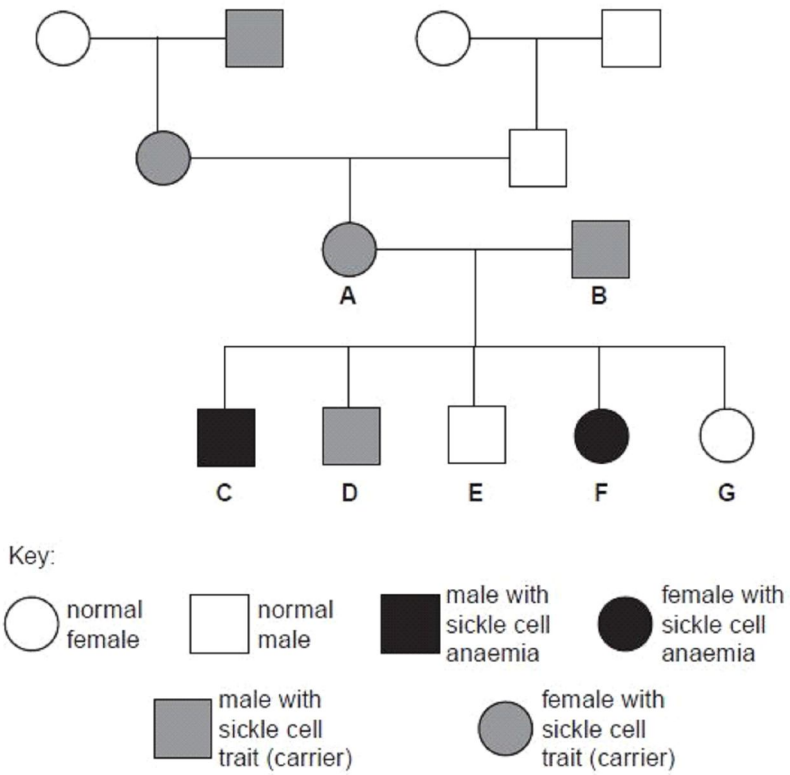


Fig. 4.1

(a) Using the symbols H^N for the allele for normal haemoglobin and H^S for the allele for sickle cell haemoglobin, state the genotypes of the following individuals.

A .....

C .....

[1]

(b) Draw a genetic diagram to show the probability of the parents **A** and **B** producing another child with sickle cell anaemia.

[4]

[Total : 5]

Q22.

2 A maize plant produced a total of 381 grains, 216 purple and smooth, 79 purple and shrunken, 65 yellow and smooth and 21 yellow and shrunken.

(a) Using the symbols **A** for purple and **a** for yellow and **B** for smooth and **b** for shrunken, draw a genetic diagram to explain these results.

[4]

(b) Explain why yellow shrunken grains breed true.

.....

.....

.....[2]

A chi-squared test was carried out to test the significance of the differences between the observed and expected results.

Table 2.1

grain phenotype	observed number	observed ratio	expected ratio	expected number	[obs no. – exp no.]^2 ÷ expected no.
purple and smooth	216	10.3	9	381 × 9/16 = 214	4/214 = 0.019
purple and shrunken	79	3.8	3	381 × 3/16 = 71	64/71 = 0.901
yellow and smooth	65	3.1	3	.....	.....
yellow and shrunken	21	1.0	1	.....	.....
total number	381			chi square value	.....

(c) Complete the missing spaces in the Table 2.1 [3]

Table 2.2

	probability greater than						
degrees of freedom	0.50	0.20	0.10	0.05	0.02	0.01	0.001
3	2.37	4.64	6.25	7.82	9.84	11.34	16.27

(d) Use the calculated values of chi-squared test and the table of probabilities to find the probability of the observed ratio of phenotypes differing significantly from the expected.

.....[1]

(e) State what conclusions may be drawn from the probability found in **(d)**.

.....

.....

.....[2]

[Total : 12]

Q23.

2 The colour of the tips of the hair in Australian Shepherd dogs is controlled by a gene at the **A** locus. There are three alleles at this locus which are:

$\mathbf{A^s}$ Black hair tips

$\mathbf{A^y}$ Red hair tips

$\mathbf{A^t}$ Copper hair tips

A cross between two dogs with copper hair tips will always produce offspring with copper hair tips. A cross between two dogs with black hair tips may produce some offspring with red hair tips and some with copper hair tips.

(a) State the ratio of **offspring** phenotypes from the following crosses:

(i) $\mathbf{A^sA^t \times A^yA^y}$

offspring phenotypes .....

ratio .....

(ii) $\mathbf{A^sA^t \times A^tA^y}$

offspring phenotypes .....

ratio .....

(iii) $\mathbf{A^yA^t \times A^yA^t}$

offspring phenotypes .....

ratio ..... [6]

(b) A dog breeder wishes to know whether a dog with red hair tips is either homozygous or heterozygous for this characteristic.

(i) State the cross needed to determine the dog's genotype.

.....

..... [1]

(ii) Explain why the offspring of this cross will reveal the genotype of the dog.

.....

.....

.....

.....

.....

..... [3]

[Total: 10]

Q24.

3 **(a)** Outline the symptoms of cystic fibrosis (CF).

.....

.....

.....

.....

.....

.....

..... [4]

(b) CF is caused by a recessive mutation, **b**, on an autosome.

Draw a genetic diagram to show, for parents with genotypes **BbXX** and **BbXY**, the **probability** of having a daughter who suffers from CF.

In your genetic diagram, show the genotypes of the gametes and the genotypes and phenotypes of the offspring.

genetic diagram

parental genotypes **BbXX** x **BbXY**

genotypes of gametes

genotypes and phenotypes of offspring

[4]

(c) One of the many mutations for CF results in the amino acid arginine being replaced by histidine in the polypeptide encoded by the CF gene.

Explain how a mutation may cause such a change in the amino acid sequence of a polypeptide.

.....

.....

.....

.....

.....

.....

..... [4]

(d) A genetic test was performed on two individuals, **D** and **E**, to find the base sequences of a small part of the CF gene. The different base sequences are shown diagramatically in Fig. 3.1. Individual **E** has CF.

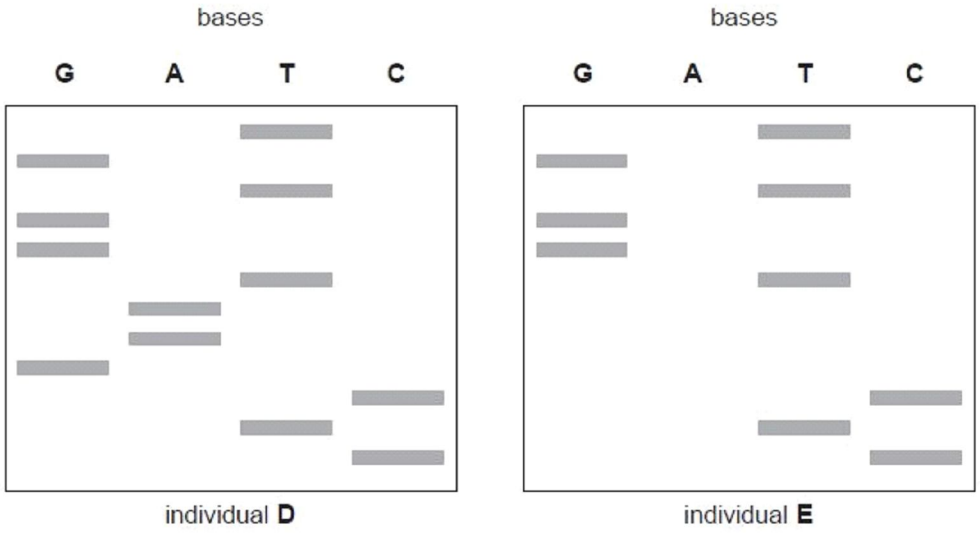


Fig. 3.1

With reference to Fig. 3.1, state,

(i) how the base sequence of **E** differs from that of **D**

.....

..... [1]

(ii) the effect of this difference in the polypeptide produced by the two individuals.

.....

.....

.....

..... [2]

[Total: 15]

Q25.

8 In mice there are several alleles of the gene that controls the intensity of pigmentation of the fur.

The alleles are listed below in order of dominance with **C** as the most dominant.

C = full colour
$\mathbf{C}^{ch}$ = chinchilla
$\mathbf{C}^{h}$ = himalayan
$\mathbf{C}^{p}$ = platinum
$\mathbf{C}^{a}$ = albino

The gene for eye colour has two alleles. The allele for black eyes, **B**, is dominant, while the allele for red eyes, **b**, is recessive.

A mouse with full colour and black eyes was crossed with a himalayan mouse with black eyes. One of the offspring was albino with red eyes.

Using the symbols above, draw a genetic diagram to show the genotypes and phenotypes of the offspring of this cross.

[6]

[Total: 6]

26.

7 **(a)** The fruit fly, *Drosophila melanogaster*, feeds on sugars found in damaged fruits. A fly with normal features is called a wild type. It has a striped body and its wings are longer than its abdomen. There are mutant variations such as an ebony coloured body or vestigial wings. These three types of fly are shown in Fig. 7.1.

wild type **ebony body** **vestigial wing**

Fig. 7.1

Wild type features are coded for by dominant alleles, **A** for wild type body and **B** for wild type wings.

Explain what is meant by the terms *allele* and *dominant*.

allele .....

.....

dominant .....

.....[2]

(b) Two wild type fruit flies were crossed. Each had alleles **A** and **B** and carried alleles for ebony body and vestigial wings.

Draw a genetic diagram to show the possible offspring of this cross.

[6]

(c) When the two heterozygous fruit flies in **(b)** were crossed, 384 eggs hatched and developed into adult flies.

A chi-squared (χ^2) test was carried out to test the significance of the differences between observed and expected results.

$$\chi^2 = \Sigma \frac{(O - E)^2}{E}$$

where Σ = sum of
O = observed value
E = expected value

(i) Complete the missing values in Table 7.1.

Table 7.1

	phenotypes of *Drosophila melanogaster*			
	grey body long wing	grey body vestigial wing	ebony body long wing	ebony body vestigial wing
observed number (O)	207	79	68	30
expected ratio	9	3	3	1
expected number (E)	216	72	72	24
O – E	-9	.....	-4	6
$(O - E)^2$	81	.....	16	36
$\frac{(O - E)^2}{E}$	0.38	.....	0.22	1.50

[3]

Table 7.2 relates χ^2 values to probability values.

As four classes of data were counted the number of degrees of freedom was 4 – 1 = 3. Table 7.2 gives values of χ^2 where there are three degrees of freedom.

Table 7.2

probability greater than	*0.50*	*0.20*	*0.10*	*0.05*	*0.01*	*0.001*
values for χ^2	2.37	4.64	6.25	7.82	11.34	16.27

(iii) Using your value for χ^2, and Table 7.2, explain whether or not the observed results were significantly different from the expected results.

.....

.....

.....

.....[2]

[Total: 14]

Q27.

7 Pompe disease is a rare neuromuscular disease caused by an autosomal recessive allele. This allele prevents the production of an enzyme called acid alpha-glucosidase (AG), which breaks down glycogen in muscle cells. Glycogen can build up in muscle cells causing damage to the cells. This damage leads to muscle weakness which gets worse with time.

(a) Explain how two parents, both of whom produce normal amounts of AG, can produce a child with Pompe disease.

.....

.....

.....

.....

.....

.....[3]

(b) One form of treatment is enzyme replacement therapy where AG is given through regular injections.

(i) Suggest how AG may be manufactured.

.....

.....[1]

(ii) Name the hormone that stimulates the breakdown of glycogen in **liver** cells.

.....[1]

(iii) State under what conditions glycogen would need to be broken down in liver or muscle cells.

.....

.....[1]

(c) The MN blood group system is based on the presence of glycoproteins M and N, on the surface membrane of red blood cells, which act as *antigens*.

State what is meant by the term *antigen*.

.....

.....[1]

(d) The type of MN antigen on the surface membrane of red blood cells is controlled by a single gene with two alleles, L^M and L^N. The phenotypes of the MN blood group system are MM, MN and NN.

Complete the genetic diagram to show how the MN blood group is inherited.

parental phenotypes MN x MN

parental genotypes ....

gametes .....

offspring genotypes .....

offspring phenotypes ..... [3]

(e) Allele frequencies for L^M and L^N vary in different human populations throughout the world.

Table 7.1 shows the L^M and L^N allele frequencies from five populations.

Table 7.1

population	allele frequency / %	
	L^M	L^N
Canadian Inuit	91	9
Egyptian	52	48
German	55	45
Chinese	57	43
Nigerian	55	45

Discuss the data shown in Table 7.1.

.....

.....

.....

.....

.....

.....[3]

[Total: 13]

Q28.

6 In sickle cell anaemia the recessive allele Hb^S replaces the normal allele Hb^A.

- The frequency of Hb^S is much higher in West Africa than in most parts of the world.
- The frequency of Hb^S corresponds with the distribution of malaria.

(a) Explain what is meant by the term *allele*.

.....

.....[1]

(b) State whether the likely life expectancy is high or low in West Africa for individuals with the following genotypes. In each case give a reason for your answer.

Hb^AHb^A .....

.....

Hb^AHb^S .....

.....

Hb^SHb^S .....

.....[4]

(c) Explain why populations of West African descent living in the USA have a decreased frequency of the Hb^S allele compared to West African populations.

.....

.....

.....

..... [2]

[Total: 7]

Q29.

9 Tuberous Sclerosis Complex (TSC) is a genetic condition caused by a dominant allele of the *TSC* gene, which leads to abnormal growth of tissue in organs such as the heart, lungs and kidneys.

Children with TSC can, with treatment, lead reasonably normal lives.

About 33% of people with TSC have at least one parent with the condition.

(a) Explain the meaning of the terms *dominant* and *gene*.

dominant .....

.....

gene .....

.....

.....[2]

(b) A couple wish to start a family. The man does not have TSC but the woman does have TSC. The woman's father does not have the condition.

Complete the genetic diagram below to show the probability of the couple's first child having TSC.

key
TSC allele = T
normal allele = t

parental phenotypes	man without TSC	woman with TSC
parental genotypes	.....	.....
gametes	.....	.....
offspring genotypes	.....	
offspring phenotypes	.....	

probability of first child having TSC ..... [3]

(c) Suggest how a person may develop TSC when there is no family history of the condition.

.....

.....

.....

.....

.....[2]

[Total: 7]

Q30.

6 A group of plants, known as Rapid Cycling Brassicas (RCBs), has been developed for use in schools and colleges for genetics experiments.

When RCB seedlings develop they can have either purple stems or non-purple stems. Their seed leaves can be either green or yellow-green.

Purple stems and green seed leaves are controlled by dominant alleles.

The genes for stem colour and seed-leaf colour are located on separate chromosomes.

(a) Explain what is meant by a *dominant allele*.

allele .....

.....

dominant .....

.....[2]

(b) Draw a genetic diagram to show the likely outcome of a cross between two RCB plants which are heterozygous for **both** stem colour and seed-leaf colour.

Use the symbols **A / a** for stem colour and **B / b** for seed leaf colour.

[6]

[Total: 8]

Q31.

6 In mice, fur colour is controlled by a gene with multiple alleles. These alleles are listed below in no particular order.

black and tan = C^{bt}	yellow = C^{y}
agouti = C^{a}	black = C^{b}

(a) Suggest explanations for the results of the following crosses between mice.

(i) Mice with agouti fur crossed with mice with black fur may produce all agouti offspring **or** some agouti and some black offspring.

.....

.....

.....

.....[2]

(ii) Crosses between heterozygous parents with the genotype $C^{y}\ C^{b}$ always produce a ratio of two yellow mice to one black mouse.

.....

.....

.....

.....[2]

(iii) Mice with yellow fur crossed with mice with black fur will produce one of the following outcomes:

- some yellow offspring and some agouti offspring
- some yellow offspring and some black and tan offspring
- some yellow offspring and some black offspring.

.....

.....

.....

.....[2]

(b) A test cross is used to determine the genotype of an organism.

Describe how you would carry out a test cross to determine the genotype of a black and tan mouse.

.....

.....

.....

.....

.....

.....[2]

[Total: 8]

Q32.

1 **(a)** Huntington's disease (HD) is an inherited disease of the central nervous system. The symptoms of HD usually develop in adulthood and include uncontrollable muscular movements, short-term memory loss and changes in mood.

HD is caused by a dominant allele of the *huntingtin* gene on chromosome 4.

Explain what is meant by the terms *allele* and *dominant*.

allele .....

.....

dominant .....

.....[2]

(b) The dominant allele of the *huntingtin* gene contains many repeats of a triplet sequence of nucleotides, CAG. The age at which symptoms of HD first appear is linked with the number of CAG repeats.

This is shown in Fig. 1.1.

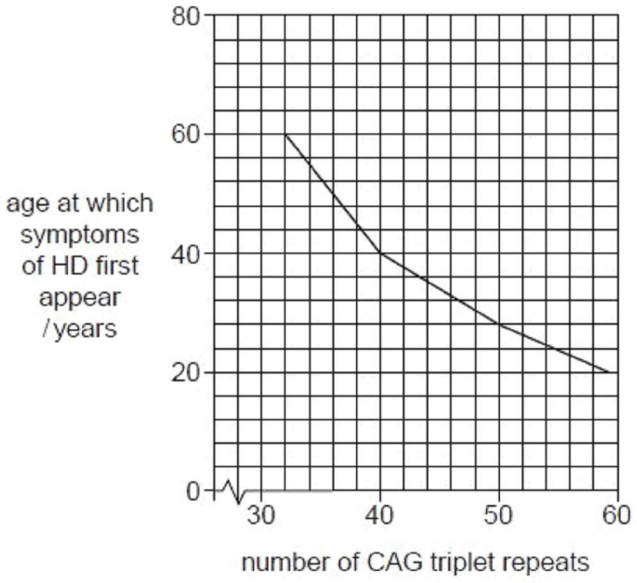


Fig. 1.1

Describe the pattern shown in Fig. 1.1.

.....

.....

.....

.....

.....[2]

(c) A blood test to detect the dominant allele is available for people at risk of HD.

Suggest why some people at risk of HD may decide **not** to take the blood test.

.....

.....

.....

.....

.....

.....

.....[3]

[Total: 7]

Q33.

1 A mutation in a gene in the fruit fly, *Drosophila melanogaster*, gives rise to white-eyed flies instead of the normal red-eyed flies. The allele for red eyes (**R**) is dominant to the allele for white eyes (**r**).

A student crossed a red-eyed fly with a white-eyed fly.

The results are shown in Table 1.1.

Table 1.1

phenotype of fly	number of offspring
red-eyed female	54
red-eyed male	0
white-eyed female	0
white-eyed male	46

(a) In *Drosophila*, males possess two different sex chromosomes, X and Y, as in humans.

Complete the genetic diagram below to show how the results in Table 1.1 could have been produced.

parental phenotypes	red-eyed fly	white-eyed fly
parental genotypes		
gametes		
offspring genotypes		
offspring phenotypes	red-eyed female	white-eyed male

[3]

(b) **(i)** The chi-squared (χ^2) test can be used to analyse the results in Table 1.1.

The expected ratio of red-eyed females to white-eyed males is 1:1.

Complete Table 1.2 and use this to calculate a value for chi-squared (χ^2).

$\chi^2 = \Sigma\frac{(O-E)^2}{E}$ $\quad\quad$ $v = n-1$

key

Σ = sum of
v = degrees of freedom
n = number of classes
O = observed value
E = expected value

Table 1.2

phenotype of fly	O	E	O–E	$(O-E)^2$	$\frac{(O-E)^2}{E}$
red-eyed female					
white-eyed male					

χ^2 = ...[3]

(ii) Use your calculated value of χ^2 and the table of probabilities below, to test the significance of the difference between observed and expected results.

degrees of freedom	probability			
	0.90	0.50	0.10	0.05
1	0.02	0.45	2.71	3.84
2	0.21	1.39	4.61	5.99

.....

.....

.....

.....[2]

[Total: 8]

Q34.

7 Occasionally during meiosis, homologous chromosomes fail to separate at anaphase. This is known as non-disjunction. Turner's syndrome is the most common chromosome mutation in human females. It can occur due to non-disjunction in meiosis during gametogenesis. Some resulting gametes will be missing an X chromosome.

Some forms of Turner's syndrome occur when one of the pair of X chromosomes is not missing but has become damaged. The damaged X chromosome may have been broken and re-formed so that part of its structure is lost.

Fig. 7.1 is a diagram of a normal X chromosome and two forms of 'damaged' X chromosomes, X_1 and X_2.

- In X_1, a section of the 'p' arm of the chromosome is missing. This deletion leads to reduced height of the female and abnormalities such as narrowing of the aorta.
- In X_2, a section of the 'q' arm of the chromosome is missing. This deletion leads to little or no development of the ovaries.

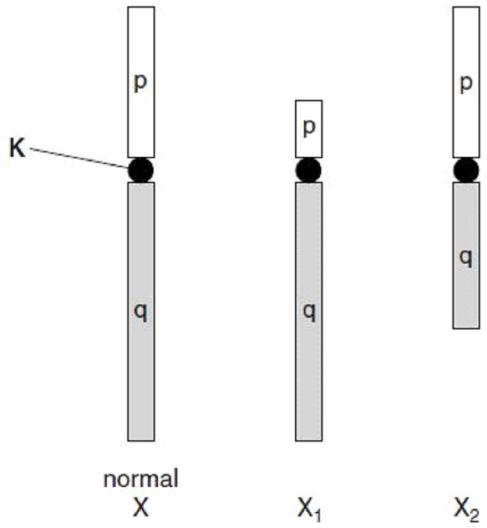


Fig. 7.1

(a) Name structure **K**.

.....[1]

(b) Explain why X_1 and X_2 result in different phenotypes.

.....

.....

.....

.....

.....

.....[2]

(c) Mothers with the X_1 form of Turner's syndrome can pass on the chromosome mutation to their daughters but not to their sons.

Complete the genetic diagram below to show how the chromosome mutation X_1 may be passed on to daughters from a mother with Turner's syndrome.

parental phenotypes	female with Turner's syndrome	normal male
parental genotypes	XX_1	
gametes		
genotypes of daughters		
phenotypes of daughters		

[4]

[Total: 7]

Q35.

7 Neurofibromatosis (NF) is a genetically inherited condition in humans where tumours grow in the nervous tissue. One symptom, which can develop around the age of 20 years, is loss of sight due to tumours on the optic nerve.

(a) If one parent has NF, there is at least a 50% chance that his or her children will develop the condition, even if the other parent is unaffected.

Complete the genetic diagram below to show how NF may be transmitted from parent to child.

key to symbols

.....

.....

parental phenotypes	parent with NF	unaffected parent
parental genotypes		
gametes		
offspring genotypes		
offspring phenotypes		

[3]

(b) Suggest how a person may develop NF when there is no family history of the condition.

.....

.....

.....

.....

.....[2]

(c) Suggest how a tumour on the optic nerve could prevent the transmission of nerve impulses to the brain.

.....

.....

.....

.....

.....

.....[3]

[Total: 8]

Q36.

7 Phenylketonuria (PKU) is a genetic disease which results in a raised concentration of the amino acid phenylalanine in the blood. If left untreated in a newborn baby, it can lead to brain damage. For this reason, babies may be tested for PKU soon after birth.

Usually, excess phenylalanine is converted to the amino acid tyrosine by the enzyme phenylalanine hydroxylase (PAH). PKU can be the result of a recessive mutation of the gene coding for PAH.

(a) Explain what is meant by a *recessive mutation*.

.....

.....

.....

.....

.....[2]

(b) Using appropriate symbols, complete the diagram below to show how two parents who do not have PKU can have children with, or without, PKU.

key to symbols

.....

.....

parental phenotypes	without PKU	without PKU
parental genotypes		
gametes		
offspring genotypes		
offspring phenotypes		

[3]

(c) PKU can be caused when a short length of the RNA produced during transcription of the gene coding for PAH is lost.

Suggest what effect this would have on the protein that is subsequently produced.

.....

.....

.....

.....

.....

.....[3]

[Total: 8]

Q37.

7 The ABO blood grouping system in humans is determined by a single gene with three alleles: I^A, I^B and I^o.

(a) Distinguish between the terms *gene* and *allele*.

gene .....

.....

.....

allele .....

.....

.....[4]

(b) Fig. 7.1 shows a pedigree diagram of the inheritance of the ABO blood group system. The blood group of some of the individuals is given in the pedigree.

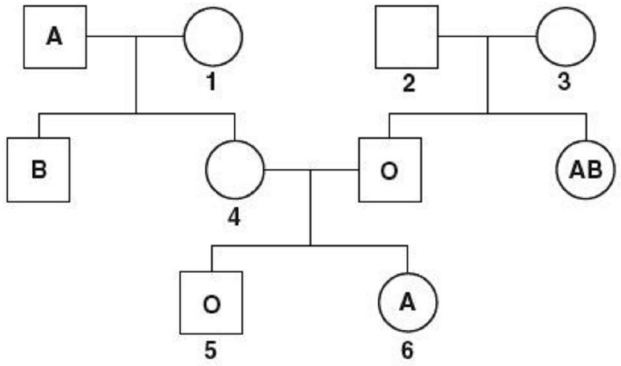


Fig. 7.1

Fig. 7.1

Use the information in Fig. 7.1 and the genetic symbols I^A, I^B and I^o, to complete the table.

individual	phenotype	genotype
1		
2		
3		
4		

[4]

[Total: 8]

Section-B

1.

9 **(a)** Outline the behaviour of **chromosomes** during meiosis. [9]

(b) Describe the ways by which **gene** mutations can occur. [6]

[Total: 15]

2.

11 **(a)** Explain how changes in the nucleotide sequence of DNA may affect the amino acid sequence in a protein. [8]

(b) Explain how the allele for haemophilia may be passed from a man to his grandchildren. You may use genetic diagrams to support your answer. [7]

[Total: 15]

3.

9 **(a)** Describe the **first** division of meiosis (meiosis I) in animal cells. [6]

(b) Discuss the link between the frequency of sickle cell anaemia and the number of cases of malaria. [9]

[Total: 15]