CAMBRIDGE INTERNATIONAL AS & A LEVEL BIOLOGY · 9700

BIOLOGY



EXAM QUESTION PACK

16 INHERITANCE

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Chapter 14

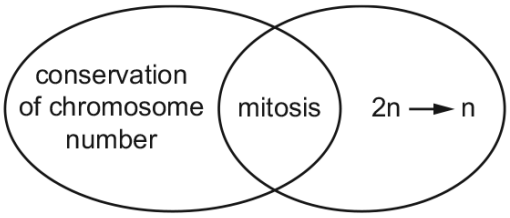
Inherited change

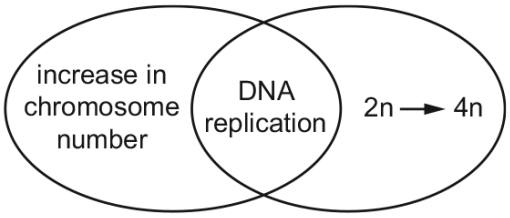
14.1 Passage of information from parent to offspring

1611. 9700_s15_qp_13 Q: 19

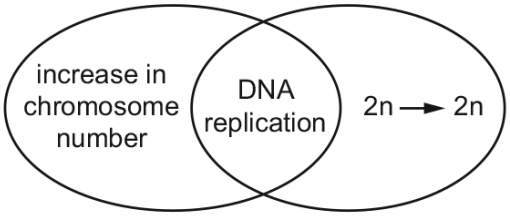
Which diagrams show the correct relationships?

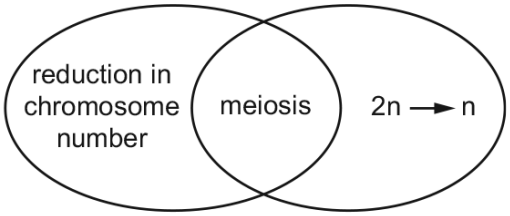
A



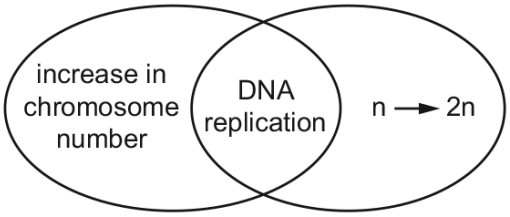


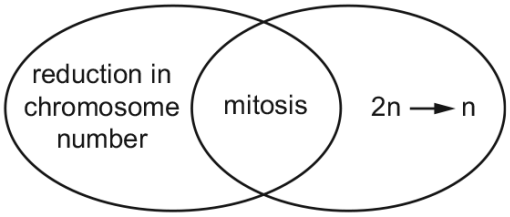
B



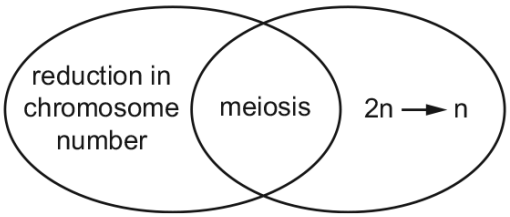


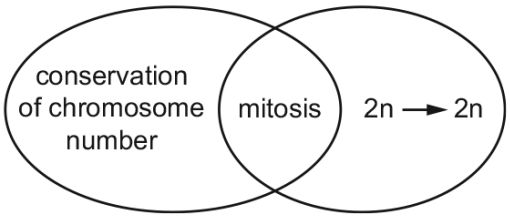
C





D

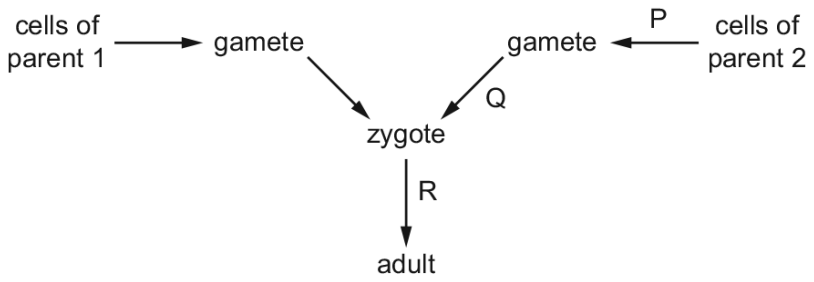




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1612. 9700_w15_qp_11 Q: 19

The diagram represents stages in the life cycle of an organism.



Which arrow represents a process that results in haploid cells and which represents mitosis?

	produces haploid cells	mitosis
A	P	Q
B	Q	R
C	R	Q
D	P	R

1613. 9700_w15_qp_13 Q: 19

Which row shows the products of mitosis, reduction division (meiosis) and fertilisation?

	products of mitosis	products of meiosis	product of fertilisation
A	four diploid cells	two diploid cells	diploid cell
B	four haploid cells	four diploid cells	haploid cell
C	two diploid cells	four haploid cells	diploid cell
D	two diploid cells	two haploid cells	haploid cell

14.2 The roles of genes in determining the phenotype

1614. 9700_w15_qp_13 Q: 20

Which statements describe how a gene mutation can lead to the production of a non-functional protein?

1 During transcription an incorrect nucleotide is added to a DNA molecule.

2 A codon in the mRNA transcribed from the mutated gene is changed.

3 The order of the bases in an anticodon on tRNA is altered during translation.

A 1, 2 and 3 **B** 1 and 2 only **C** 2 and 3 only **D** 2 only

---

Q1.

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

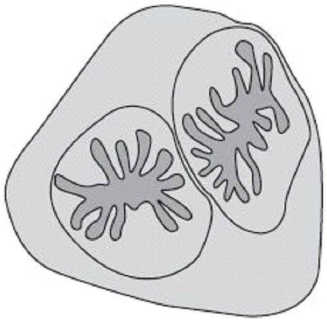


Fig. 4.1

(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.

.....

.....

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.....

.....[4]

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

.....

.....

.....

.....

.....

[4]

[Total : 10]

Q2.

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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.

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black female X orange male	orange female X black male	

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(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.

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.....[1]

[Total: 9]

Q4.

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

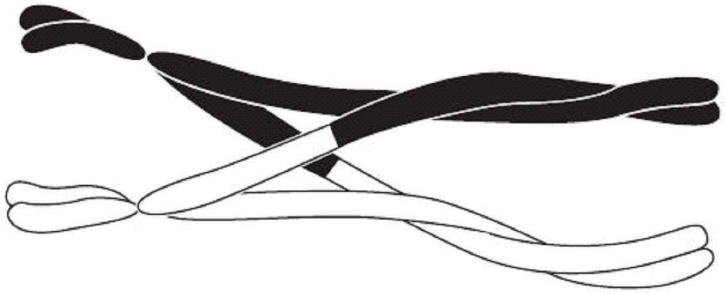


Fig. 2.1

(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.

.....

.....

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.....

.....

.....[4]

[Total: 10]

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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.

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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.

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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]

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(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.

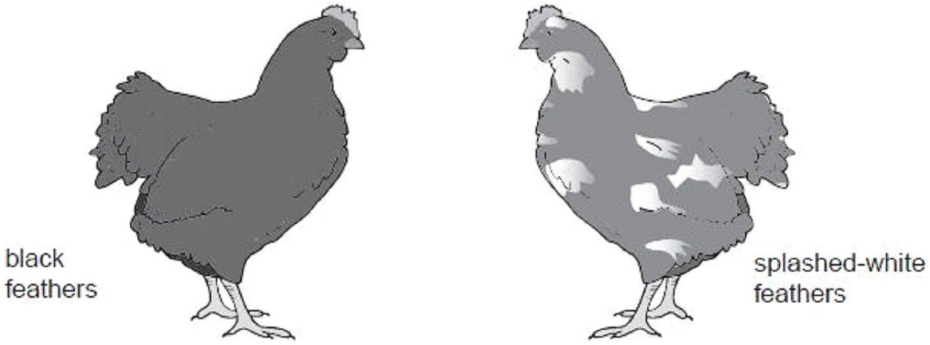


Fig. 7.1

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.

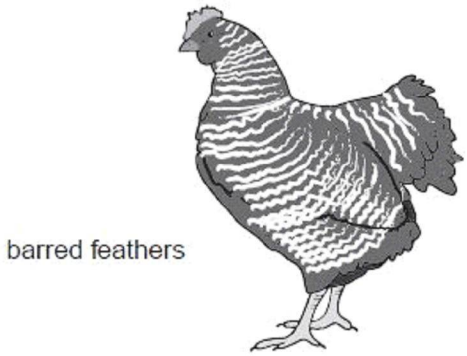


Fig. 7.2

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(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.

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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.

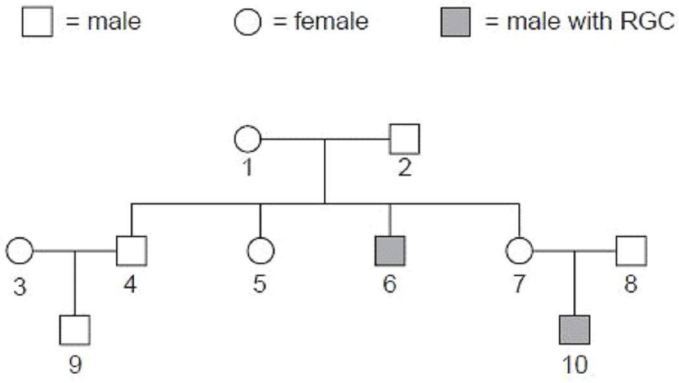


Fig. 6.1

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(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

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[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.

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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.

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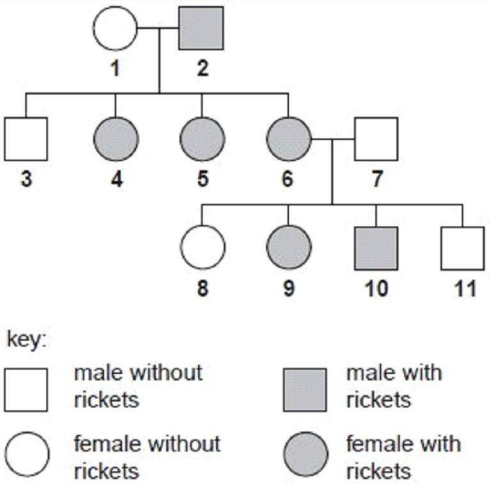


Fig. 6.1

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]

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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.

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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

.....

.....

parental phenotypes	unaffected man	unaffected woman
parental genotypes		
gametes		
offspring genotypes		
offspring phenotypes		

[3]

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

.....

.....

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.....

.....

.....

.....

..... [3]

[Total: 8]

Q12.

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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.

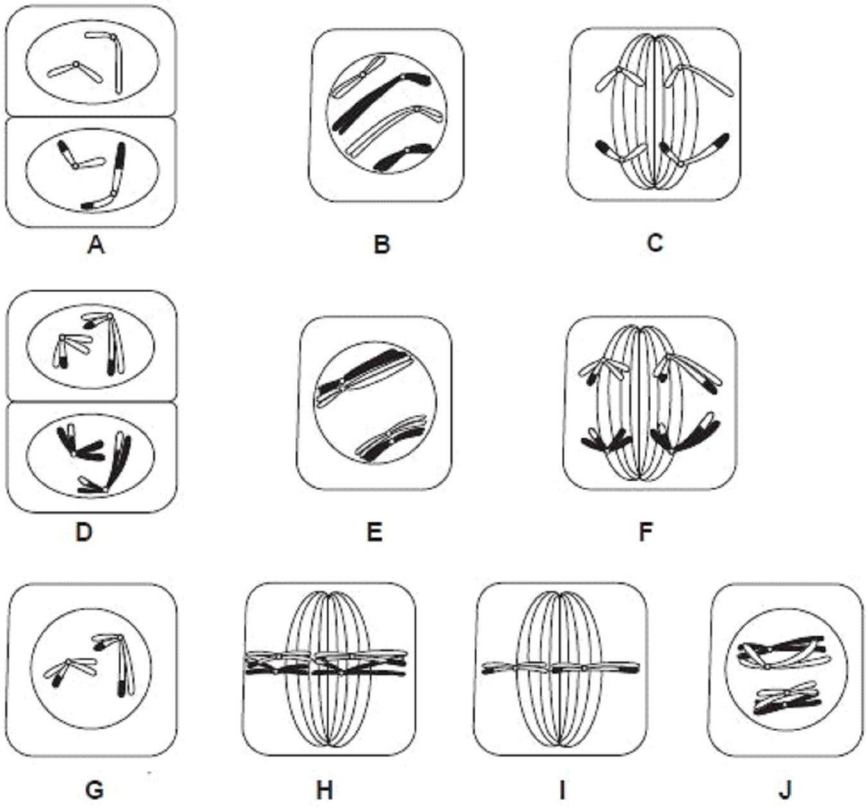


Fig. 7.1

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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.

.....

.....

.....

.....

.....

.....

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.....

.....

.....[5]

[Total: 9]

Q13.

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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.

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

(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]

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(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]

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(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.

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[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 .....

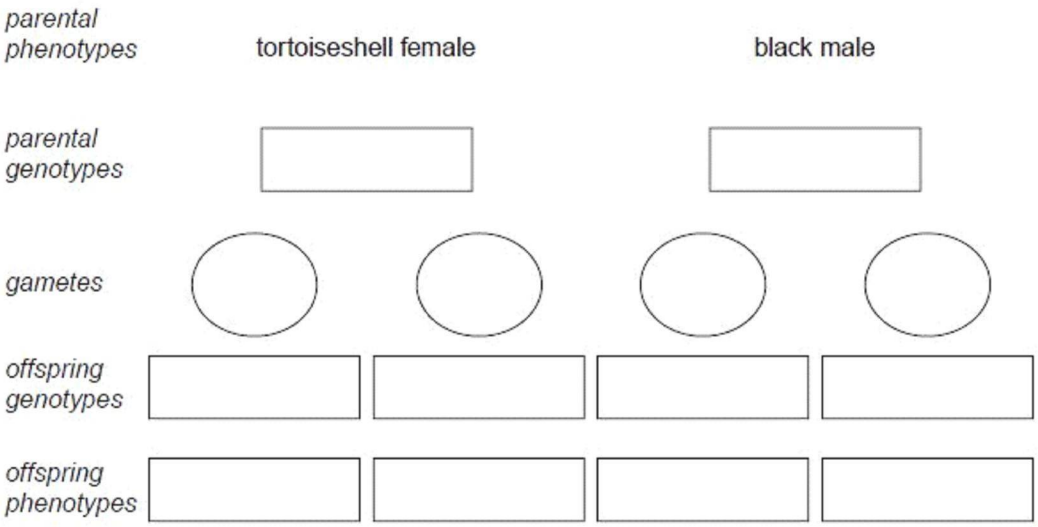
.....

gene .....

..... [2]

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(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.

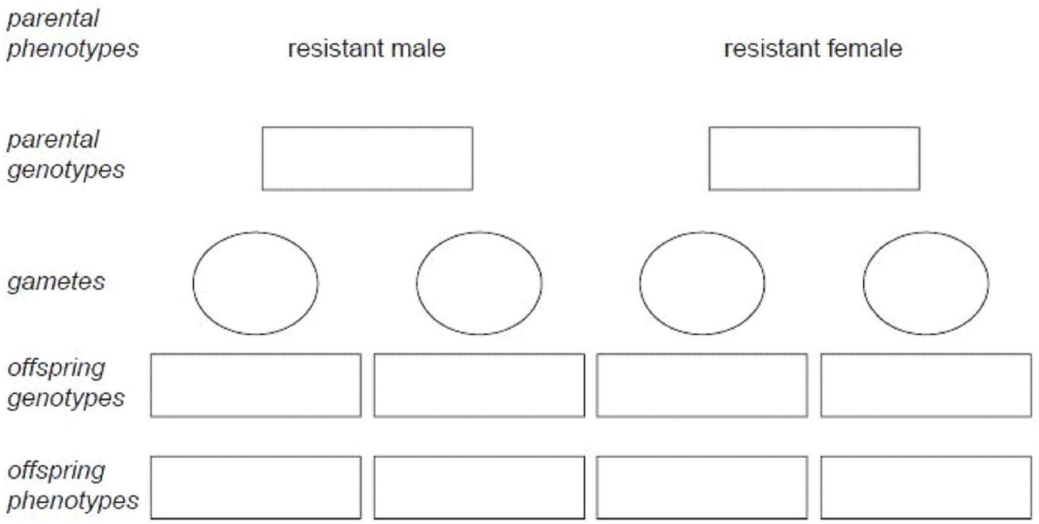
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(a) Using appropriate symbols, complete the genetic diagram to show how two resistant rats can produce warfarin-susceptible offspring.

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]

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(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.

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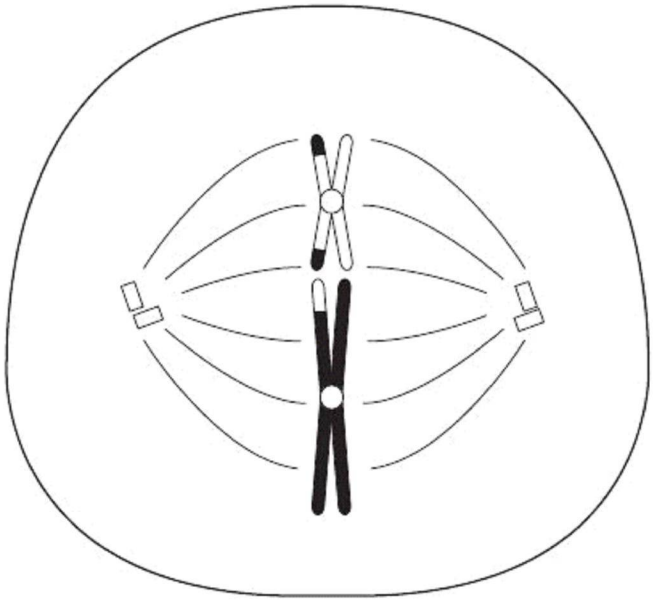
.....

..... [3]

[Total: 9]

Q18.

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



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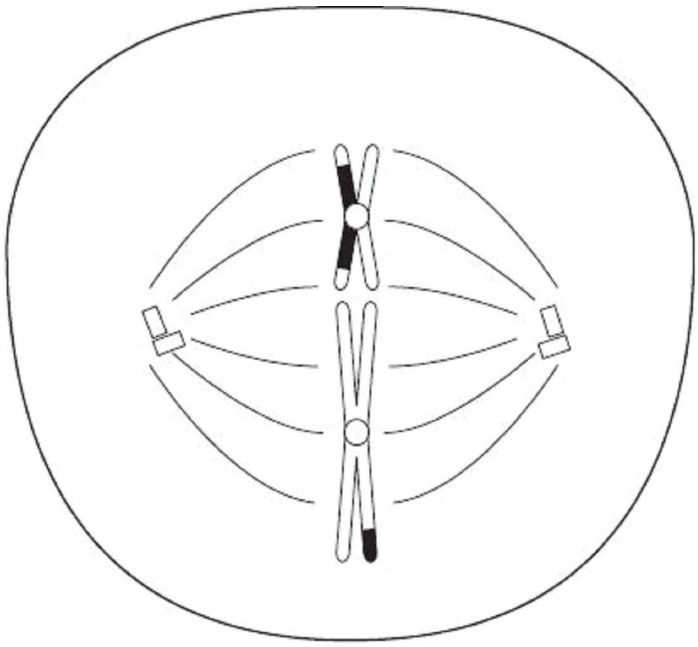


Fig. 4.1

(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]

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(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.

.....

.....

.....

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[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.

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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]

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(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.

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

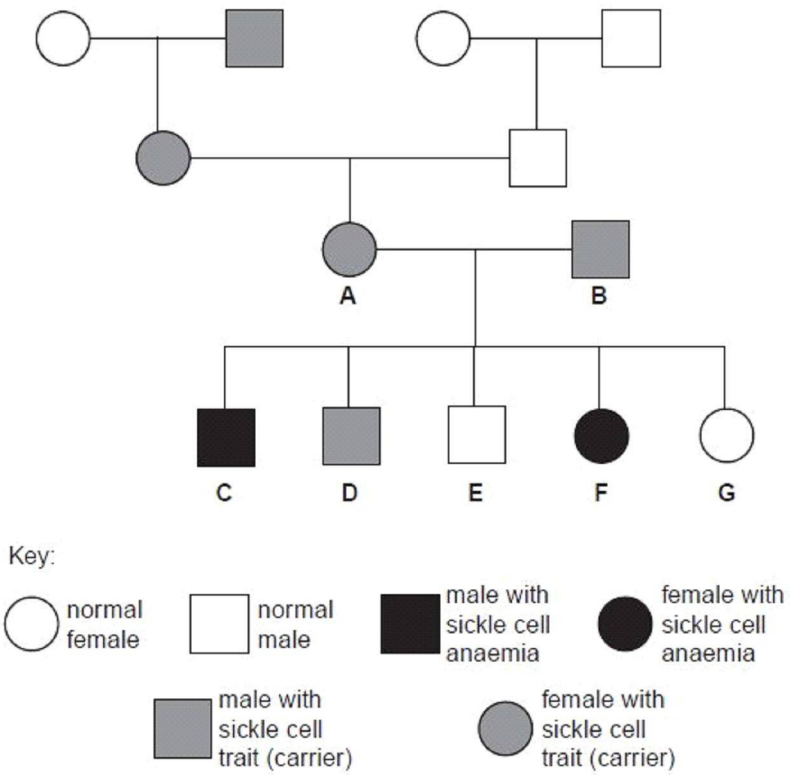


Fig. 4.1

(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.

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[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.

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[4]

(b) Explain why yellow shrunken grains breed true.

.....

.....

.....[2]

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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.

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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]

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(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.

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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]

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(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.

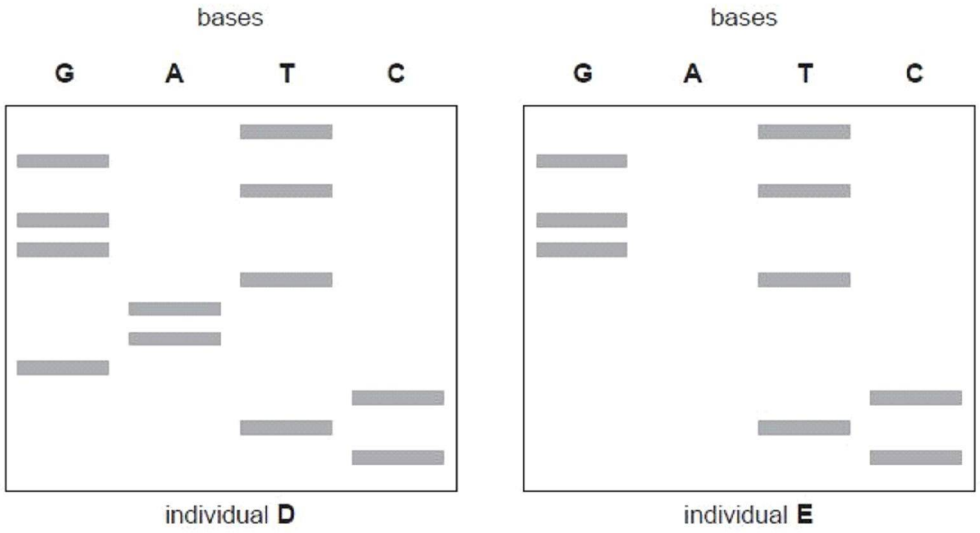


Fig. 3.1

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.

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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.

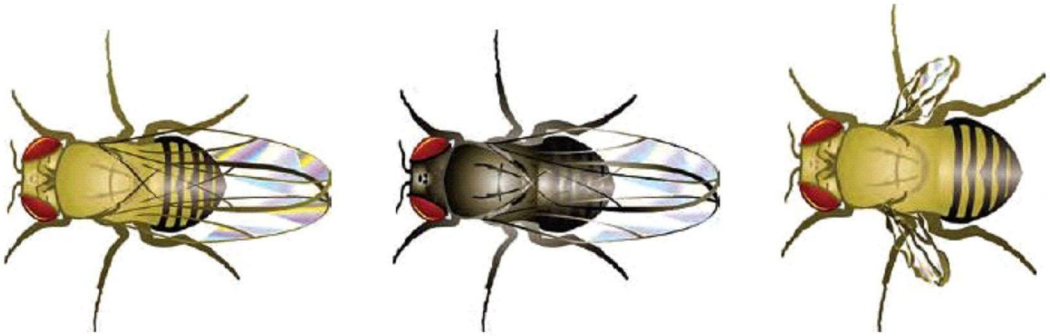
[6]

[Total: 6]

26.

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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.

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[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

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(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]

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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]

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(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

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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]

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(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]

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(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.

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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.

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[6]

[Total: 8]

Q31.

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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]

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(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.

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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.

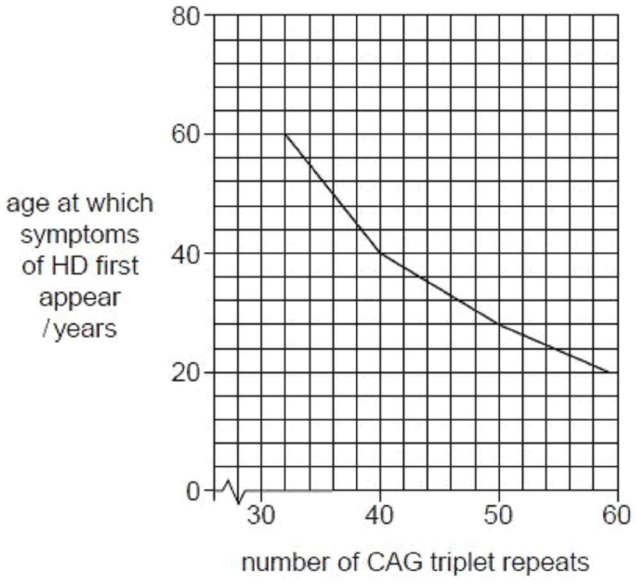


Fig. 1.1

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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

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(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).

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

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]

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(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.

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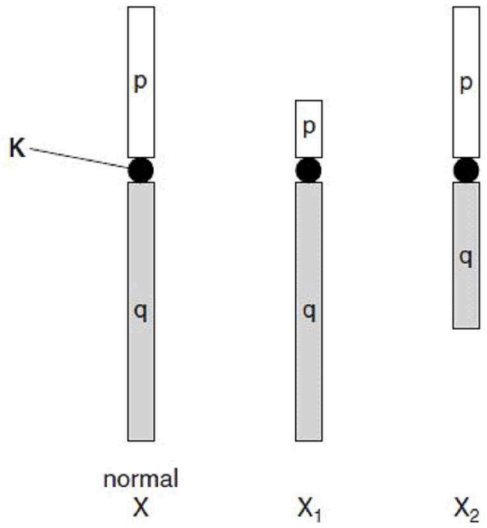


Fig. 7.1

(a) Name structure **K**.

.....[1]

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

.....

.....

.....

.....

.....

.....[2]

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(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.

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(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]

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(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]

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(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.

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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.

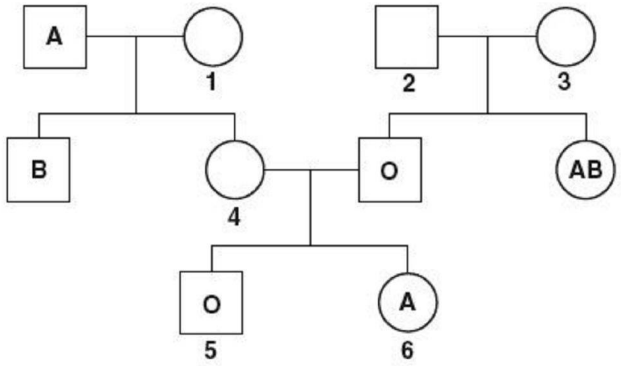


Fig. 7.1

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]

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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]

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1. Nov/2018/Paper_41/No.2

In the fruit fly, *Drosophila melanogaster*, two different genes control body colour and eye colour.

- **G/g** are alleles of the body colour gene.
- **G** results in grey body, **g** results in black body.

- **R/r** are alleles of the eye colour gene.
- **R** results in red eyes, **r** results in brown eyes.

Each gene is autosomal.

A dihybrid cross was carried out using a fly with a grey body and red eyes crossed with a fly with a black body and brown eyes. Both parents were homozygous for both genes. The offspring from the F1 generation were crossed to obtain the F2 offspring.

(a) A statistical test showed that the results of the cross were significantly different from those expected.

State the name of the statistical test used **and** state the expected phenotypic ratio for the F2 generation.

statistical test .....

expected ratio .....

[2]

(b) A test cross can be carried out in order to identify flies from an F2 generation that are heterozygous for both genes.

Draw a genetic diagram to show how a test cross between a heterozygous grey-bodied, red-eyed F2 fly and a fly with a black body and brown eyes can produce four different offspring phenotypes.

Use the symbols **G/g** and **R/r**.

[4]

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(c) The results of the test cross in **(b)** are shown in Table 2.1. These results are significantly different from the expected results.

Table 2.1

phenotypes of offspring of test cross	number of individuals
grey body, red eyes	123
grey body, brown eyes	7
black body, red eyes	6
black body, brown eyes	132

Describe how these results are different from the expected results **and** explain why they are different.

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..... [5]

[Total: 11]

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1. Nov/2019/Paper_41/No.4

(a) Meiosis has an important role in sexual reproduction. Meiosis occurs during gametogenesis in humans and during the formation of pollen grains and embryo sacs in plants.

(i) Complete Table 4.1 to compare meiosis with mitosis.

For both columns, put a tick (✓) in the box if the statement is correct and put a cross (✗) in the box if the statement is incorrect.

Each box must contain either a tick or a cross.

Table 4.1

statement	meiosis	mitosis
chromosome number is maintained		
homologous chromosomes pair up		
sister chromatids separate		
occurs in prokaryotes		

[2]

(ii) Compare the role of meiosis in gametogenesis to produce sperm cells in humans with the role of meiosis in gametogenesis in producing pollen grains in flowering plants.

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..... [4]

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(b) Flower colour is important in sexual reproduction of insect-pollinated plants.

In the rosy periwinkle, *Catharanthus roseus*, flower colour is controlled by three genes, **R/r**, **D/d** and **P/p**, which interact together to control flower colour.

Fig. 4.1 is a drawing of a rosy periwinkle.

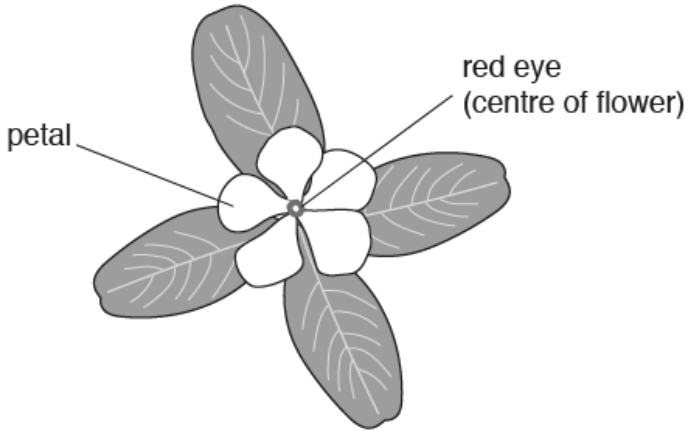


Fig. 4.1

The presence of the **R** allele results in a red pigment in the centre of the flower (red eye).

The **D** allele and the **P** allele are only expressed when the **R** allele is present.

- When the **D** allele and the **R** allele are present, the flower has dark pink petals with a red eye.
- When the **P** allele and the **R** allele are present, the flower has pale pink petals with a red eye.
- When the **D** allele, the **P** allele and the **R** allele are all present, the flower has dark pink petals with a red eye.
- The recessive alleles **r**, **d** and **p** result in no pigments being produced and the flower has white petals and no red eye.

(i) Deduce the phenotypes of these rosy periwinkle genotypes.

RR dd PP .....

Rr Dd Pp .....

rr Dd Pp .....

RR dd pp ..... [4]

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(ii) The pigments causing flower colour in the rosy periwinkle are formed by a biosynthetic pathway.

The **R** allele mutated to produce the **r** allele.

The **r** allele codes for a non-functional protein.

Explain how the mutation that changes **R** to **r** results in no red pigment being synthesised in the flower of rosy periwinkle.

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..... [4]

(iii) One mutation that changes the flower colour of rosy periwinkle occurs in a region of the DNA that does not code for a polypeptide.

Suggest what function this region of DNA might perform.

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..... [1]

[Total: 15]

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2. Nov/2019/Paper_42/No.4

(a) Meiosis has an important role in sexual reproduction. Meiosis occurs during gametogenesis in humans and during the formation of pollen grains and embryo sacs in plants.

(i) Complete Table 4.1 to compare meiosis with mitosis.

For both columns, put a tick (✓) in the box if the statement is correct and put a cross (✗) in the box if the statement is incorrect.

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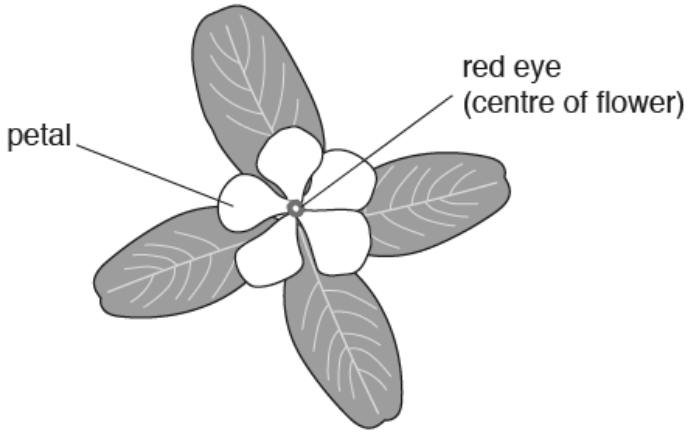


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- When the **D** allele, the **P** allele and the **R** allele are all present, the flower has dark pink petals with a red eye.
- The recessive alleles **r**, **d** and **p** result in no pigments being produced and the flower has white petals and no red eye.

(i) Deduce the phenotypes of these rosy periwinkle genotypes.

RR dd PP .....

Rr Dd Pp .....

rr Dd Pp .....

RR dd pp ..... [4]

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(ii) The pigments causing flower colour in the rosy periwinkle are formed by a biosynthetic pathway.

The **R** allele mutated to produce the **r** allele.

The **r** allele codes for a non-functional protein.

Explain how the mutation that changes **R** to **r** results in no red pigment being synthesised in the flower of rosy periwinkle.

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..... [4]

(iii) One mutation that changes the flower colour of rosy periwinkle occurs in a region of the DNA that does not code for a polypeptide.

Suggest what function this region of DNA might perform.

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..... [1]

[Total: 15]

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(a) The house mouse, *Mus musculus*, has a diploid number of 40 chromosomes.

Fig. 2.1 shows 6 of these chromosomes.



Fig. 2.1

Identify **one** pair of homologous chromosomes on Fig. 2.1 by drawing circles around **two** chromosomes. [1]

(b) Fig. 2.2 shows the banding pattern of chromosome pair 11 of *M. musculus*. The banding pattern is obtained by staining.

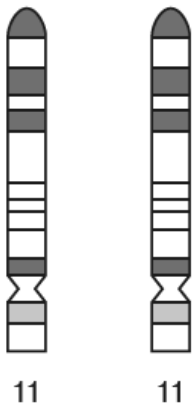


Fig. 2.2

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(i) Explain why chromosomes, such as those in Fig. 2.2, are described as a homologous pair.

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..... [3]

(ii) State the number of chromosomes that are present in *M. musculus* spermatozoa.

..... [1]

(c) *M. musculus* produces gametes by meiosis. These gametes are genetically different.

There is random fusion of gametes at fertilisation.

(i) Explain why meiosis is important in the life cycle of *M. musculus*, **apart from** producing genetically different gametes.

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..... [2]

(ii) Explain how the random fusion of gametes leads to the expression of rare, recessive alleles.

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..... [2]

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(d) A mutation causing coats of mice to be woolly in appearance is in a gene located on chromosome 11. The mutation causes a very shortened polypeptide product. Mice with the woolly coat phenotype have longer fur than mice with normal coats.

(i) Explain how a base substitution mutation can lead to a very shortened polypeptide product.

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..... [2]

(ii) The inheritance of the woolly coat characteristic was investigated.

Draw a genetic diagram to show a cross between two heterozygous parents with normal coats.

Use the symbols **A** and **a** for the alleles.

parental genotypes

gametes

offspring genotypes

offspring phenotypes

[3]

[Total: 14]

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4. Nov/2019/Paper_43/No.7(c)

(c) The height of some plants is partly controlled by their genes. Height in pea plants is affected by a gene with two alleles. The dominant allele results in the production of active gibberellin, which stimulates stem elongation.

(i) State the symbol that represents the dominant allele.

..... [1]

(ii) Explain how this dominant allele results in the production of active gibberellin.

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..... [2]

(iii) Active gibberellin stimulates stem elongation by causing the breakdown of DELLA protein repressors so that growth genes can be expressed.

Suggest the effects of the expression of these growth genes.

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..... [2]

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5. Nov/2019/Paper_43/No.10(b)

(b) Outline the effects of mutant alleles on the phenotype in Huntington's disease. [8]

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6. June/2019/Paper_41/No.2

The interpupillary distance (IPD) is the distance in millimetres between the centres of the pupils of the eyes. Fig. 2.1 shows how IPD is measured.

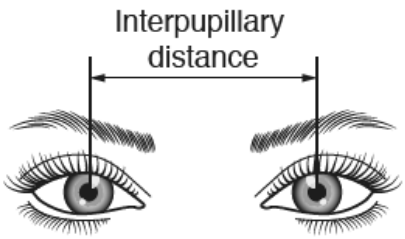


Fig. 2.1

IPD is one example of a characteristic of human facial structure that shows variation.

Fig. 2.2 shows the pattern of variation in IPD in a large sample of adults.

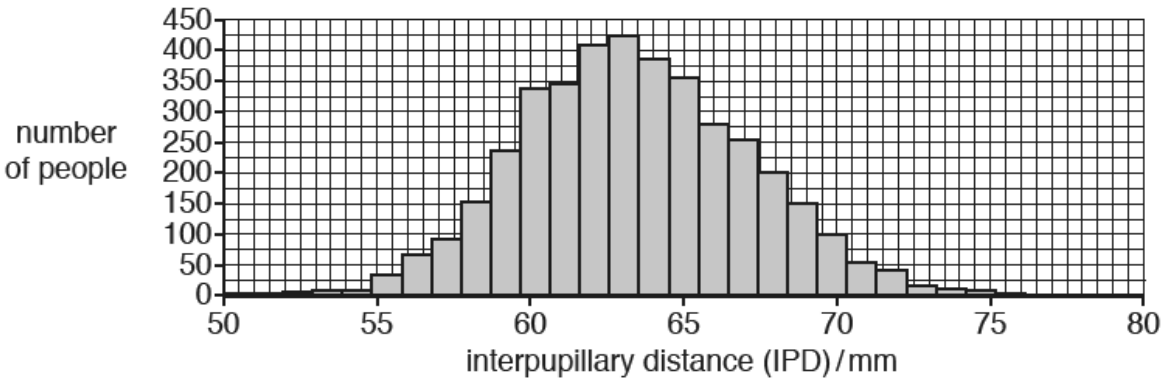


Fig. 2.2

(a) (i) Name the type of variation shown in Fig. 2.2.

..... [1]

(ii) Suggest **and** explain how genes and the environment contribute to variation in IPD in humans.

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..... [3]

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(b) Individuals with an IPD of 70 mm or more have a mutation in the *PAX3* gene that results in less PAX3 protein being made.

The normal role of the PAX3 protein is to increase the expression of many other genes involved in embryonic development. These genes affect a range of phenotypic features such as facial structure, hearing and eye colour.

(i) State the term that is used to describe a gene, such as *PAX3*, that controls the expression of other genes **and** suggest how the PAX3 protein controls the expression of other genes.

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..... [3]

(ii) Describe how microarray analysis could be used to identify the genes switched on by PAX3 in embryonic cells.

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..... [5]

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(iii) The chimpanzee, *Pan troglodytes*, has DNA that is 98.5% similar to humans, including possession of the *PAX3* gene. Investigations show that chimpanzees express higher levels of the PAX3 protein during embryonic development than humans.

Fig. 2.3 shows a chimpanzee, *Pan troglodytes*.



Fig. 2.3

Suggest how knowledge of the *PAX3* gene helps scientists explain how humans and chimpanzees are very different in facial structure, even though they have very similar DNA.

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..... [3]

[Total: 15]

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7. June/2019/Paper_41/No.3(a)

Some neurones in the brain produce a neurotransmitter known as dopamine. Parkinson's disease occurs when the neurones that produce dopamine die. A person with the disease may experience difficulty in coordinating movement, often seen as tremors (shaking) in different parts of the body.

Parkinson's disease typically occurs in people older than 55 years. Younger people with these symptoms are said to have early onset Parkinson's disease (EOPD).

Recessive mutations in a gene known as *PINK1*, located on chromosome 1, an autosome, are believed to be one cause of EOPD. A person with this form of EOPD has a homozygous recessive genotype.

(a) Draw a genetic diagram of a cross between two individuals who are heterozygous at the *PINK1* gene locus.

key to symbols used for alleles

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parental genotypes

gametes

offspring genotypes

ratio of offspring phenotypes

[4]

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8. June/2019/Paper_41/No.9(b)

(b) Describe the behaviour of chromosomes during meiosis. [8]

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9. June/2019/Paper_42/No.4

Oculocutaneous albinism (OCA) is a type of albinism. There are many different forms of OCA.

OCA1A is one form of OCA, caused by a recessive mutation in the autosomal gene, *TYR*, coding for the enzyme tyrosinase. This enzyme is involved in the biosynthetic pathway that results in the production of melanin, the pigment responsible for the colour of hair, skin and eyes.

A person with OCA1A has white hair, very pale skin and pink eye colour.

(a) Draw a genetic diagram to show the probability of a child having OCA1A, if both parents are carriers.

Use the symbols **A** and **a** for the alleles.

parental genotypes

gametes

offspring genotypes

offspring phenotypes

probability

[4]

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(b) Fig. 4.1 shows the biosynthetic pathway involving tyrosinase.

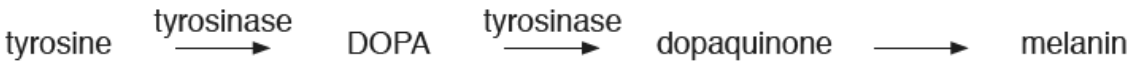


Fig. 4.1

There are a number of different mutations of the *TYR* gene that can result in an absence of melanin and cause OCA1A. These include:

- a missense mutation, caused by a base substitution, is most common
- a nonsense mutation, caused by a base substitution, is less common
- an insertion mutation, which is extremely rare.

(i) A missense mutation results in a complete polypeptide chain that does not fold properly to form the functioning enzyme.

A nonsense mutation results in a shortened polypeptide.

Explain why a missense mutation results in a different product from a nonsense mutation.

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..... [2]

(ii) Explain how an insertion mutation in *TYR* can lead to a lack of melanin in a person with OCA1A.

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..... [4]

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(c) Worldwide, 1 in 17 000 people are born with OCA. This compares with 1 in 165 people among the Guna people of Panama. The Guna people of Panama have a small population and mostly live on many small islands off the coast of Panama.

Suggest reasons why the Guna population of Panama has a relatively high number of cases of OCA.

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..... [2]

[Total: 12]

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10. June/2019/Paper_43/No.2

Albinism is a condition that results from disruption of a biosynthetic pathway involving the enzyme tyrosinase. It is estimated that 1 in 17 000 people in the world has albinism.

(a) A recessive mutation in the *TYR* gene, which codes for the enzyme tyrosinase, is one cause of albinism. Individuals with this form of albinism are homozygous recessive.

Describe the phenotype of a person with albinism.

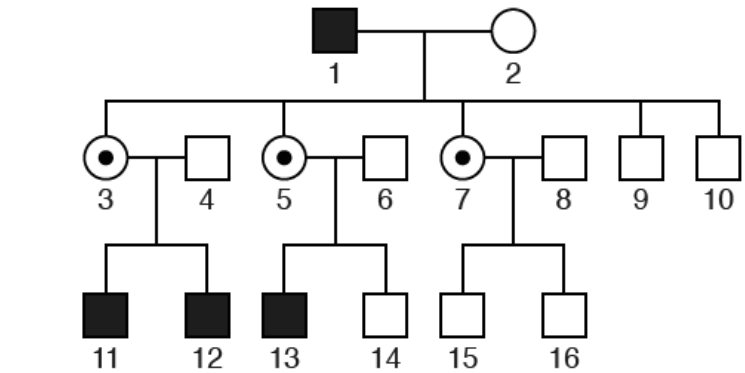
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..... [2]

(b) A recessive mutation in a different gene causes a type of albinism that mainly affects the eyes (ocular albinism). A person with this condition has reduced clarity of vision and involuntary eye movements.

Fig. 2.1 shows the pattern of inheritance of ocular albinism in one family. The pattern indicates sex-linked inheritance.



Key:

= unaffected female

= carrier female

= unaffected male

= male with ocular albinism

Fig. 2.1

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(i) Explain why Fig. 2.1 supports sex-linked inheritance of ocular albinism.

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..... [3]

(ii) Draw a genetic diagram to show how individuals 1 and 2 **cannot** have a child with ocular albinism.

key to symbols

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parental genotypes

gametes

offspring genotypes

offspring phenotypes

[4]

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(iii) Ocular albinism may be caused by a base deletion mutation. This mutation results in a non-functional protein.

Explain how a base deletion mutation can result in a non-functional protein.

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..... [2]

(iv) Ocular albinism is a non-progressive disorder and clarity of vision remains stable throughout life.

A female has a family history of ocular albinism but she does not have any symptoms. A test to find out if she has the mutant allele is available.

Suggest **one** reason for taking this test and **one** reason against taking this test.

for .....

.....

against .....

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[2]

[Total: 13]

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11. Feb/2019/Paper_42/No.6(a)

MELAS syndrome is an inherited disease caused by a mutation in a gene located in mitochondrial DNA (mtDNA). All mtDNA is inherited from the mother.

Fig. 6.1 shows four generations of a family where several individuals are affected by MELAS syndrome.

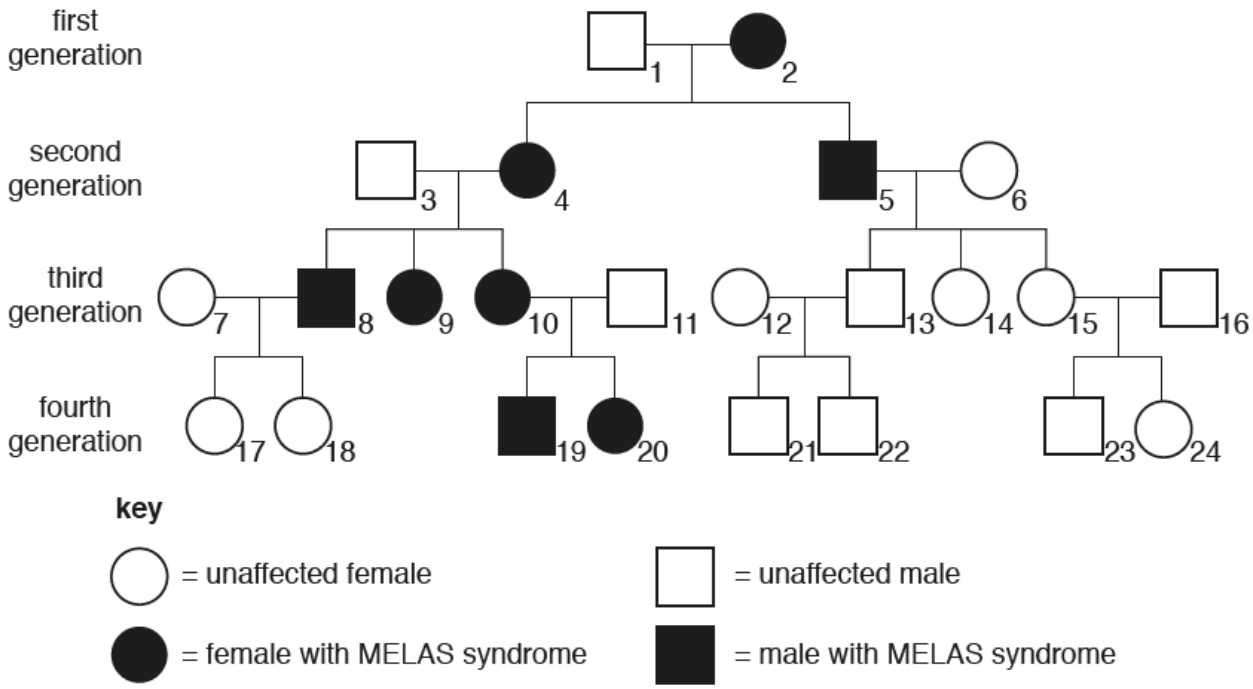


Fig. 6.1

(a) With reference to Fig. 6.1, state **and** explain the evidence that MELAS syndrome is an mtDNA disease and **not** a disease caused by a mutation in a gene on the X chromosome.

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..... [4]

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12. Feb/2019/Paper_42/No.10(b)

(b) Explain how meiosis **and** fertilisation can result in genetic variation amongst offspring. [8]

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1. Nov/2021/Paper_41/No.6

Fig. 6.1 outlines the first three stages of respiration in aerobic conditions.

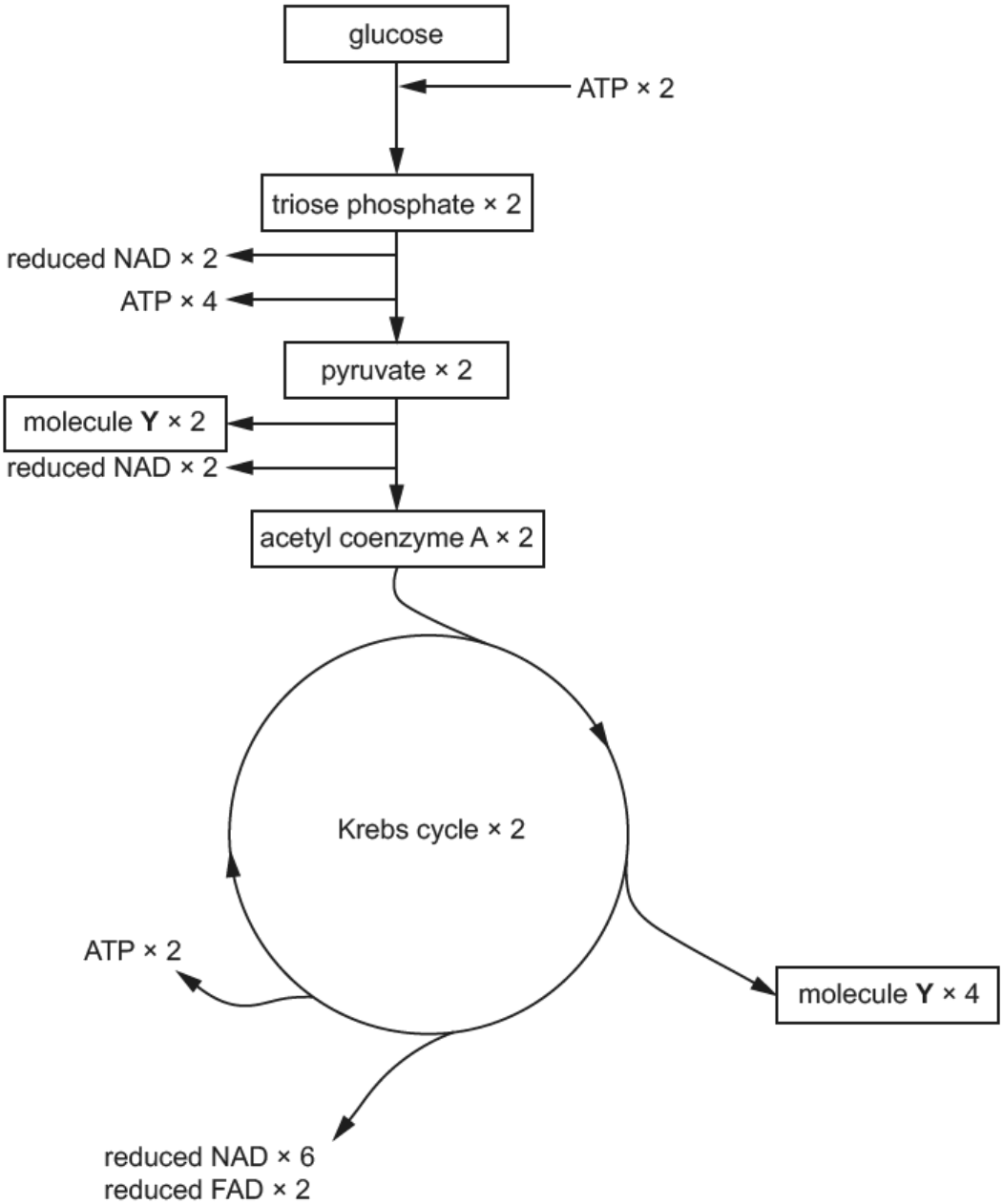


Fig. 6.1

(a) Name molecule **Y** in Fig. 6.1.

..... [1]

(b) Explain how Fig. 6.1 shows that glycolysis involves oxidation.

.....

..... [1]

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(c) At one time it was thought that the oxidative phosphorylation of:

- one molecule of reduced NAD results in the synthesis of 3 ATP molecules
- one molecule of reduced FAD results in the synthesis of 2 ATP molecules.

Using Fig. 6.1, a theoretical value for the net number of ATP molecules that are synthesised for each molecule of glucose can be calculated.

Modern research has shown that the actual net number of ATP molecules synthesised for each glucose molecule respired is much lower than this theoretical value.

(i) Using Fig. 6.1, calculate the theoretical value for the net number of ATP molecules that are synthesised for each molecule of glucose respired in **all** phosphorylation reactions.

Show your working.

answer = ... [2]

(ii) Suggest **two** reasons why the actual net number of ATP molecules synthesised is less than the theoretical number.

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..... [2]

(d) Outline the roles of NAD and FAD in aerobic respiration.

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..... [3]

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(e) Rotenone is used as an insecticide. Rotenone kills insects by inhibiting the transfer of electrons in the electron transport chain of the mitochondrion.

Explain how rotenone affects ATP synthesis in the mitochondrion.

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..... [3]

[Total: 12]

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1. June/2022/Paper_41/No.7

(a) The fruit fly, *Drosophila melanogaster*, usually has red eyes. A gene for eye colour has four alleles: red, apricot, honey and white.

Define the terms *gene* and *allele*.

gene .....

.....

allele .....

.....

[2]

(b)
- The allele for red eyes, C^R, is dominant to the other three alleles.
- The allele for apricot eyes, C^A, is dominant to the allele for honey eyes, C^H.
- The allele for white eyes, C^W, is recessive to the other three alleles.

Construct a genetic diagram to show the genotypes, phenotypes and ratio of the offspring from a cross between a fruit fly with red eyes, C^RC^H, and a fruit fly with apricot eyes, C^AC^W.

parents phenotypes	red eyes	apricot eyes
parents genotypes	C^RC^H	C^AC^W
gametes		
offspring genotypes		
offspring phenotypes		
ratio		

[3]

(c) Describe how you would carry out a test cross **and** use it to determine the genotype of a red-eyed fruit fly.

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..... [3]

[Total: 8]

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2. June/2022/Paper_42/No.1

The jaguar, *Panthera onca*, is a large cat that lives mainly in South America. The majority of jaguars have light brown fur with black spots, as shown in Fig. 1.1. Some jaguars have completely black fur, as shown in Fig. 1.2.



Fig. 1.1



Fig. 1.2

(a) The pigments involved in fur colour are produced as a result of biochemical pathways that take place in cells called melanocytes. These pathways are similar to those that occur in human melanocytes.

The melanocortin 1 receptor (MC1R) is located on the cell surface membrane of melanocytes and is coded for by the *MC1R* gene.

Fig. 1.3 outlines the processes that occur in jaguar melanocytes.

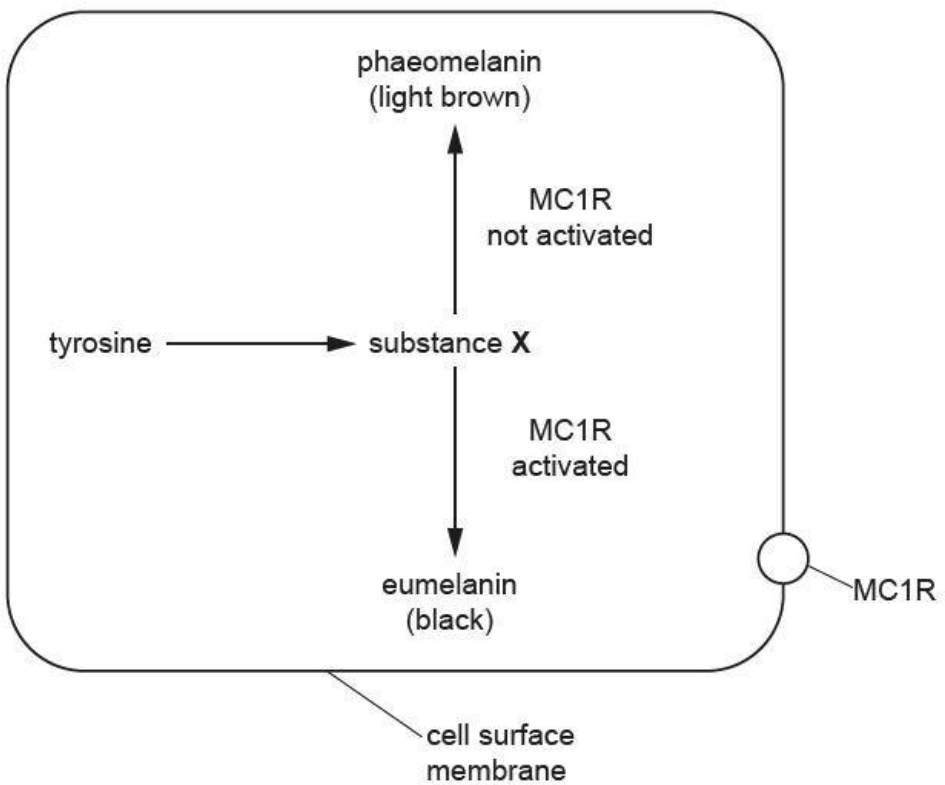


Fig. 1.3

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(i) Name the substance represented by **X**.

..... [1]

(ii) When MC1R is activated a second messenger is produced in the cell.

Give an example of a second messenger.

..... [1]

(iii) Substance **X** is also produced in humans, but a mutation of the *TYR* gene can result in substance **X** not being produced.

Describe the phenotype of a person with this mutation.

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..... [2]

(b) The *MC1R* gene has two alleles and is located on an autosome.

- When two jaguars with light brown fur mate all the offspring have light brown fur.
- When two jaguars with black fur mate either all the offspring will have black fur or some offspring will have black fur and some will have light brown fur.

Using symbols, construct a genetic diagram to show how two jaguars with black fur can produce some offspring with black fur and some offspring with light brown fur.

[4]

[Total: 8]

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3. June/2022/Paper_43/No.7

(a) Epistasis occurs when a gene at one locus can affect the expression of a gene at another locus.

Define the terms gene and locus.

gene .....

.....

locus .....

.....

[2]

(b) Fur colour in mice, *Mus musculus*, is determined by a number of genes. One example is the result of epistatic interaction between two genes, **A** and **B**.

- Allele **A** codes for the production of pigment in the fur.
- Allele **a** does not code for the production of pigment and results in white fur (albino).

- Allele **B** codes for the production of brown fur.
- Allele **b** codes for the production of black fur.

Construct a genetic diagram to show the results, including the ratio, of a cross between two mice heterozygous for both genes.

parent genotypes **AaBb** **x** **AaBb**

parent phenotypes

gametes

ratio..... [6]

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1. Nov/2022/Paper_41/No.1

Adenosine deaminase (ADA) deficiency is an immune system disorder caused by a recessive autosomal mutation.

Severe combined immunodeficiency caused by a lack of ADA is called ADA-SCID.

(a) Genetic engineering is used to make a recombinant human protein to treat people with ADA-SCID.

Outline the principles of genetic engineering.

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..... [4]

(b) In 2016 gene therapy to cure ADA-SCID was approved in Europe. The gene therapy involves three main steps.

- Blood (haematopoietic) stem cells are taken from the bone marrow of the person with ADA-SCID.
- The functional gene and its promoter are inserted into the blood stem cells.
- A single infusion (injection) of the gene-corrected cells is given to the patient.

(i) Explain why a single infusion of gene-corrected stem cells is enough to cure the disease.

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..... [2]

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(ii) Explain why a promoter has to be transferred as well as the desired gene.

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..... [2]

(iii) A modified retrovirus is used to insert the new gene into the DNA of the blood stem cells.

State **two** ethical considerations of using a retrovirus for gene therapy.

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..... [2]

(c) The gene therapy technique used to cure ADA-SCID is **not** suitable for treating the genetic disease called Huntington's disease. A newer technique called gene editing could potentially be used instead to cure Huntington's disease.

Explain why gene editing is more suitable as a potential cure for Huntington's disease.

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..... [3]

[Total: 13]

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2. June/2022/Paper_42/No.1

(a) In some varieties of domestic cats the gene for fur colour is located on the **X** chromosome. This gene has two alleles. One allele codes for black fur and the other allele codes for ginger fur. The two alleles are codominant so a heterozygous cat will have fur with patches of black and ginger colours. A cat with fur of two colours is known as a tortoiseshell.

Using appropriate symbols, construct a genetic diagram to show the results of a cross between a female tortoiseshell cat and a male ginger cat.

symbols

parent phenotypes tortoiseshell female ginger male

parent genotypes

gametes

offspring genotypes

offspring phenotypes

[5]

(b) In humans the *TYR* gene is involved in the production of a dark pigment, melanin, in some cells.

Describe how the expression of the *TYR* gene leads to the production of melanin.

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..... [3]

[Total: 8]

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3. June/2022/Paper_43/No.7

The determination of sex in domestic turkeys is different from that in humans. The sex chromosomes in turkeys are named **Z** and **W**. Male turkeys are **ZZ** and female turkeys are **ZW**.

The gene for feather colour is located only on the **Z** chromosome.

- The dominant allele codes for bronze feathers.
- The recessive allele codes for brown feathers.

(a) Define the terms dominant and recessive.

dominant .....

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recessive .....

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[2]

(b) Using suitable symbols, construct a genetic diagram to show the results of a cross between a heterozygous bronze male turkey and a brown female turkey.

symbols

parent phenotypes bronze male brown female

parent genotypes

gametes

offspring genotypes

offspring phenotypes

[5]

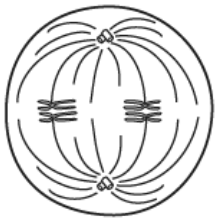
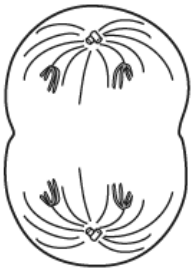
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1. March/2023/Paper_ 9700/42/No.5

Meiosis is described as a reduction division because the number of chromosomes in the daughter cells is reduced by half.

(a) Table 5.1 describes some of the events that take place during four of the different stages of meiosis in an animal cell.

Table 5.1

stage of meiosis	spindle fibres	diagram
metaphase I	attach to centromeres and arrange homologous pairs of chromosomes at the equator of the cell	
anaphase I		
	re-form spindle in daughter cells	
telophase II	disassemble	

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Complete Table 5.1 by:

- outlining the behaviour of the spindle fibres during anaphase I
- identifying the stage of meiosis in which spindle fibres re-form the spindle in daughter cells
- drawing a diagram to show telophase II.

You do **not** need to add labels to your diagram showing telophase II.

[4]

(b) Explain the need for a reduction division during meiosis.

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[Total: 8]

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2. June/2023/Paper_ 9700/41/No.6

Domestic rabbits vary in the length and colour of their fur.

Fig. 6.1 shows a domestic rabbit with short fur and a fur colour pattern called Himalayan.

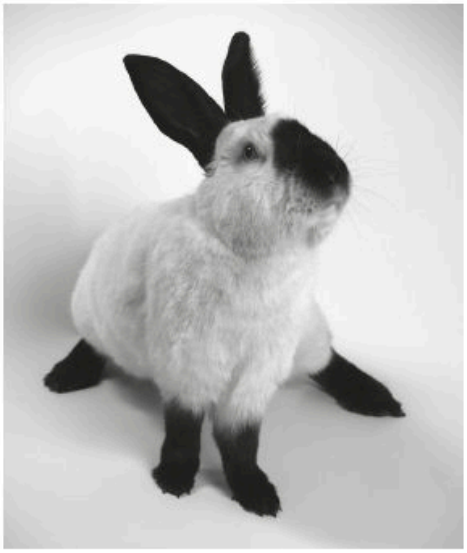


Fig. 6.1

The two genes that determine the length and colour of the fur of this rabbit occur at the **A / a** locus and the **B / b^h / b** locus. These two gene loci are on separate chromosomes.

- The allele **A** results in short fur.
- The allele **a** results in long fur.
- **A** is dominant to **a**.

- The allele **B** results in black fur all over the body.
- The allele **b^h** results in black fur on the nose, ears, paws and tail of the rabbit, and white fur on the rest of the body (Himalayan pattern).
- The allele **b** results in white fur all over the body (albino).
- **B** is dominant to **b^h** and **b^h** is dominant to **b**.

(a) (i) List the **four** possible genotypes of the rabbit shown in Fig. 6.1.

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..... [2]

(ii) One phenotype of rabbit always breeds true. This means that when it is mated to a rabbit that looks the same as itself, all the offspring look the same as the parents.

Describe the **phenotype** of the rabbit that breeds true.

..... [1]

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(b) A rabbit with long, black fur all over the body that was homozygous at both loci was crossed with a rabbit with short, white fur that was homozygous at both loci. The F1 offspring had short, black fur. These F1 rabbits were mated together to become the parents of the F2 generation.

Draw a genetic diagram to predict the F2 offspring genotypes and the ratio of F2 offspring phenotypes.

F1 genotypes:

gametes:

F2 offspring genotypes:

ratio of F2 offspring phenotypes:

[5]

(c) A rabbit breeder performed multiple crosses of the type described in **(b)**. This gave enough data to test the prediction that the genes for fur length and fur colour show independent assortment.

(i) Explain why the two genes assort independently.

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..... [3]

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(ii) The rabbit breeder placed the results in a table and started to calculate the χ^2 value.

Table 6.1 shows the results and some of the calculations made.

Table 6.1

phenotype	**observed number (*O*)**	**expected number (*E*)**	$(O - E)^2$	$\frac{(O - E)^2}{E}$
short, black fur	25		4	0.148
long, black fur	11		4	0.444
short, white fur	8		1	0.111
long, white fur	4		1	0.333

Calculate the expected numbers and write them in the shaded column in Table 6.1. [1]

(iii) Use the formula provided and the figures in Table 6.1 to calculate the χ^2 value.

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

χ^2 = ... [1]

[Total: 13]

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3. June/2023/Paper_ 9700/41/No.8

Fig. 8.1 shows a cell from the testis of a locust at the late prophase I stage of meiosis.



Fig. 8.1

Explain how the behaviour of the chromosomes in prophase I of meiosis results in the appearance shown in Fig. 8.1.

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[Total: 4]

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4. June/2023/Paper_ 9700/41/No.10

Sweet peas are garden plants that vary in height.

- Tall sweet peas grow to 200 cm in height.
- Dwarf sweet peas grow to 30 cm in height.
- Tall sweet peas contain a dominant ***Le*** allele.
- Dwarf sweet peas are homozygous for the recessive ***le*** allele.

Explain how the ***lele*** genotype results in the dwarf phenotype in sweet peas, with reference to the effect of ***lele*** on:

- enzyme synthesis
- hormone production
- the expression of genes affecting plant growth.

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[Total: 6]

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5. June/2023/Paper_ 9700/42/No.2

(a) In sexual reproduction, meiosis occurs to produce haploid gametes from a diploid cell.

Explain the meaning of the terms haploid **and** diploid.

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..... [2]

(b) Describe the behaviour of **chromosomes** during the main stages of meiosis I in animal cells.

prophase I .....

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metaphase I .....

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anaphase I .....

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telophase I .....

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[4]

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(c) The production of haploid gametes by meiosis also involves division of the cytoplasm.

State the term used to describe this division of the cytoplasm.

..... [1]

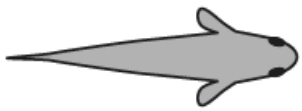
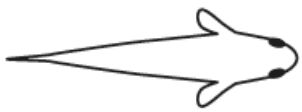
[Total: 7]

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6. June/2023/Paper_ 9700/43/No.5

A freshwater fish species, *Oryzias latipes*, has individuals with four body colour patterns, as shown in Table 5.1.

Table 5.1

phenotype	body colour pattern
red	
white	
red with black spots	
white with black spots	

Two unlinked genes determine the body colour patterns shown in Table 5.1.

One gene controls whether the body colour is red or white:
- dominant allele **R** = red
- recessive allele **r** = white.

The other gene controls whether black spots are present or **not** present:
- dominant allele **B** = with black spots
- recessive allele **b** = without black spots.

A fish that is homozygous recessive at both loci is white.

Genetic crosses were carried out to investigate the inheritance of the four different body colour patterns.

Males that were red with black spots, and homozygous at both loci, were crossed with females that were white. The F1 offspring were all red with black spots.

These F1 offspring were then crossed to produce the F2 generation.

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(a) Table 5.2 shows the observed numbers obtained of each of the four different phenotypes for the F2 generation.

Table 5.2

phenotype	observed	expected	$O–E$	$(O–E)^2$	$\frac{(O–E)^2}{E}$
red with black spots	279	281.25			
white with black spots	95	93.75	1.25	1.5625	0.017
red	96	93.75	2.25	5.0625	0.054
white	30	31.25			
				χ^2 = ...	

Table 5.2 compares the observed numbers with the numbers that would be expected in the F2 generation for a normal dihybrid ratio.

Calculate χ^2 for the F2 generation by completing Table 5.2.

The formula for χ^2 is:

$$\chi^2 = \sum \frac{(O–E)^2}{E}$$

[3]

(b) The critical value at $p = 0.05$ and 3 degrees of freedom is 7.815.

Comment on whether the null hypothesis should be accepted or rejected.

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..... [2]

Further analysis of the results from the F2 generation in Table 5.2 showed that there were no white males or white males with black spots.

In *O. latipes*, females have two **X** chromosomes and males have an **X** and a **Y** chromosome.

It was deduced that, in *O. latipes*:

- the gene that controls body colour is located on the **X** chromosome **and** the **Y** chromosome
- the gene that controls whether black spots are present or **not** is located on an autosome.

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(c) To produce the F2 generation, red males with black spots, X^rY^RBb, were crossed with red females with black spots, X^RX^rBb.

Complete the Punnett square in Fig. 5.1 to show the genotypes and phenotypes of the F2 generation.

- Use the symbols X^R, X^r and Y^R for the alleles of the gene that controls body colour.
- Use the symbols **B** and **b** for the alleles of the gene that controls whether black spots are present or **not**.

Some of Fig. 5.1 has been completed for you.

	female gametes			
male gametes	X^RB	X^rB	X^Rb	X^rb
X^rB	X^RX^rBB		X^RX^rBb	X^rX^rBb
	female		female	female
	red + black spots		red + black spots	white + black spots
Y^RB	X^RY^RBB	X^rY^RBB	X^RY^RBb	X^rY^RBb
	male	male	male	male
	red + black spots	red + black spots	red + black spots	red + black spots
X^rb	X^RX^rBb	X^rX^rBb	X^RX^rbb	
	female	female	female	
	red + black spots	white + black spots	red + no spots	
Y^Rb		X^rY^RBb		X^rY^Rbb
		male		male
		red + black spots		red + no spots

Fig. 5.1 [4]

(d) Explain why there are no white males or males that are white with black spots in the F2 generation.

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..... [2]

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(e) In another cross, red males with the genotype $\mathbf{X^rY^Rbb}$ were mated with white females with the genotype $\mathbf{X^rX^rbb}$. All the male offspring were expected to be red and all the female offspring were expected to be white.

The observed results showed that the offspring included two red females out of 253 and one white male out of 198.

Suggest an explanation for this unexpected result.

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..... [2]

[Total: 13]

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1. Nov/2023/Paper_9700/41/No.6

The tiger barb, *Puntigrus tetrazona*, is a South American fish that is popular worldwide as an aquarium fish. Fig. 6.1 shows the appearance (phenotype) of a normal (wild-type) tiger barb.



Fig. 6.1

- Tiger barbs that show a wild-type phenotype are gold with black stripes.
- Tiger barbs that show an albino phenotype are gold with white stripes.
- In 2012, a fish breeder discovered a tiger barb with a new, transparent, phenotype. This fish had a transparent body and black stripes.

The fish breeder crossed the tiger barb showing the new transparent phenotype with a tiger barb showing the albino phenotype.

All the F1 offspring were wild-type. These F1 offspring were crossed with each other.

Table 6.1 shows the phenotypes obtained in the F2 generation and the number of fish showing each phenotype.

Table 6.1

F2 phenotype	number of fish
wild-type (gold with black stripes)	173
albino (gold with white stripes)	57
transparent with black stripes	58
transparent with white stripes	19

(a) **(i)** The fish breeder concluded that the new transparent phenotype had occurred because of a mutation.

Explain how the results in Table 6.1 support this conclusion.

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..... [2]

(ii) State the approximate whole-number ratio shown by the results in Table 6.1.

..... [1]

(iii) Explain what the results in Table 6.1 show about the genes and alleles that determine the wild-type, albino and the two different transparent phenotypes in tiger barbs.

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(b) The F1 tiger barbs all looked the same but the F2 offspring showed variation. The F2 offspring showed four different phenotypes.

Describe the processes that occurred during meiosis in the F1 fish that allowed this variation to occur.

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..... [4]

[Total: 11]

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2. Nov/2023/Paper_9700/42/No.5

The fruit fly, *Drosophila melanogaster*, has autosomal genes for body colour and wing shape.

Gene **B/b** is involved in the production of body colour:
- **B** = dominant allele for brown body colour
- **b** = recessive allele for black body colour.

Gene **D/d** is involved in wing shape:
- **D** = dominant allele for straight wing
- **d** = recessive allele for curved wing.

A dihybrid test cross was carried out between flies heterozygous for body colour and for wing shape and flies homozygous recessive for body colour and for wing shape.

(a) Table 5.1 shows the number of offspring of each phenotype obtained in the test cross.

Table 5.1

phenotype	observed number	expected number
brown body colour, straight wings	2843	
brown body colour, curved wings	855	
black body colour, straight wings	842	
black body colour, curved wings	2768	

Use Table 5.1 to calculate the expected number of each phenotype if the two genes are on **different** autosomes. Write your answers in the table. [1]

(b) A chi-squared (χ^2) test was carried out to compare the observed results with the results that would be expected from a dihybrid cross involving genes on different autosomes.

The value of $\chi^2 = 2097.836$.

Table 5.2 shows the critical values for the χ^2 distribution.

Table 5.2

degrees of freedom	*p* value		
	0.05	**0.01**	**0.001**
1	3.841	6.635	10.828
2	5.991	9.210	13.816
3	7.815	11.345	16.266
4	9.488	13.277	18.467

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Explain how the value of χ^2 and Table 5.2 can be used to assess the significance of the difference between the observed results and the expected numbers in Table 5.1.

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(c) Suggest explanations for the **observed** results in Table 5.1.

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..... [4]

[Total: 9]

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