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

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

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

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

(i) Consider a cross between two heterozygous individuals.

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

Use the space below for any working.

Answer = _____ % **[1]**

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

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

---

[4]

2. Fig. 6.1 shows a number of examples of inheritance.

A	An *Antirrhinum* plant with red flowers is crossed with one that has white flowers. All the offspring have pink flowers.
B	A haemophiliac man has children with a woman who is not a haemophiliac. Their daughters all carry the allele for the disease, but their sons do not have the disease.
C	Two *Salvia* plants with purple flowers are crossed. The offspring are produced in the ratio 9 purple-flowered : 3 pink-flowered : 4 white-flowered.
D	A short-haired black mouse crossed with a long-haired brown mouse produces all short-haired black offspring. Mating one of these offspring with the long-haired parent produces mice in the ratio of 1 short-haired black : 1 long-haired black : 1 short-haired brown : 1 long-haired brown.
E	Two snails with plain shells produce 34 offspring with plain shells and 12 with striped shells.

Fig. 6.1

Complete the table below, by matching each of the examples **A** to **E** to the correct explanation of their pattern of inheritance.

Explanation	Letter of example
One gene with two alleles. The alleles show codominance.	
One gene with two alleles located on an autosome (gene not sex linked). One allele is dominant and the other is recessive.	
Two genes for two different characteristics on two different chromosomes.	
A sex linked gene with a dominant and a recessive allele.	
Epistasis, where two genes interact to affect one phenotypic character.	

[5]

3(a). An infection by a strain of the bacterium *E. coli* called O157 can lead to the appearance of schistocytes in a blood sample.

Infections by this bacterium are the most common cause of acute kidney failure in children. This results in a condition called **HUS**.

Most children who develop **HUS** make a full recovery. A few children are left with permanent kidney damage.

(i) Name the short-term treatment given to children with acute kidney failure and **outline** the alternative long-term treatment options that would be available if necessary.

[5]

(ii) Suggest **one** further treatment that **HUS** patients may require to prevent them becoming anaemic. Explain your suggestion.

[3]

(b). Some forms of HUS are not associated with *E. coli* O157 infections. These are known as atypical HUS or AHUS.

Some cases of AHUS are known to be due to an inherited gene mutation.

Carefully read and then complete the following passage about the inheritance of AHUS and the role of the genetic counsellor.

Some inherited forms of AHUS are due to a _____ mutant allele.

Parents show no symptoms of the disease so, if they have a child who develops AHUS, the child must be _____ for this allele. Since neither parent shows symptoms, yet both carry the mutant allele, the allele does not show _____ linkage.

A genetic counsellor would use a _____ diagram to explain the risk of any further children either developing AHUS or being a carrier. Where unaffected parents have one child with AHUS, there would be a _____ percent chance of having a child who is a carrier.

[5]

4. Agammaglobulinemia and Vici syndrome are both genetic diseases.

Agammaglobulinemia results in a lack of mature B lymphocytes in a person's blood.

(i) Suggest and explain one symptom of agammaglobulinemia.

---

[2]

(ii) Fig. 4 shows the inheritance pattern of agammaglobulinemia in a family.

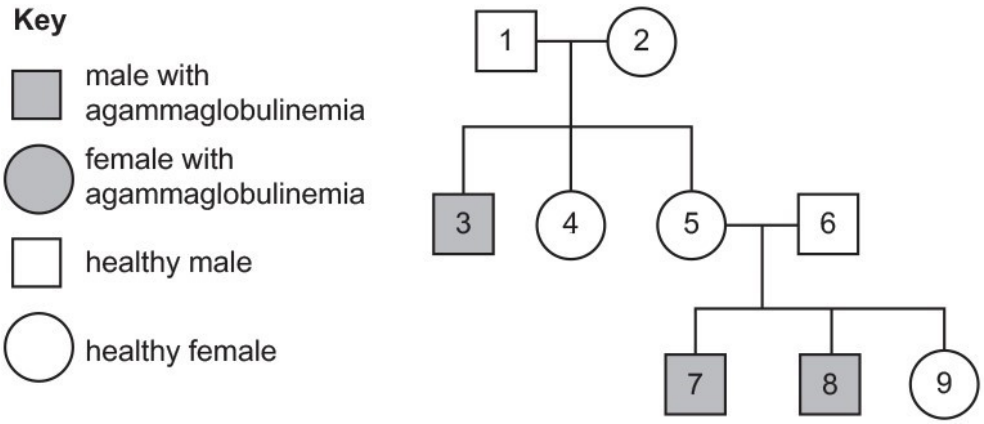


Fig. 4

What conclusions can you draw about the location and nature of the allele responsible for causing agammaglobulinemia? Explain your conclusions.

---

[4]

5. Selection pressure can affect homozygous individuals. The effect can be investigated using a model gene pool.

A large gene pool is necessary to ensure that

A genetic drift can occur if frequency is higher.
B homozygous individuals are present in high frequency.
C the effect of chance variations in gene frequencies are minimised.
D Hardy–Weinberg equilibrium is achieved.

Your answer ☐

[1]

6(a). In domesticated, farmed pigs, the following two traits have been studied:

The allele for curly tail, **T**, is dominant to the allele for straight tail, **t**.

The allele for pink skin (dermis), **D**, is dominant to the allele for black skin, **d**.

(i) Draw a genetic diagram to show the results of crossing pigs that are heterozygous for both traits, tail and skin. Use the letters given above.

parental genotypes _____ _____

gametes _____ _____

offspring phenotypes

---

phenotype ratio

---

[5]

Calculate x^2. (You may wish to use **Table 17.2** to write figures for steps in your calculation process.)

$$x^2 = \sum \frac{(O-E)^2}{E}$$

Answer_____ **[3]**

(ii) The farmer had concluded that the genes are linked.

Use your calculation and **Table 17.3** to justify whether the farmer's conclusion can be supported or not.

Degrees of freedom	Probability							
	0.95	0.90	0.75	0.50	0.25	0.10	0.05	0.01
1	0.004	0.016	0.102	0.455	1.32	2.71	3.84	6.63
2	0.103	0.211	0.575	1.386	2.77	4.61	5.99	9.21
3	0.352	0.584	1.212	2.366	4.11	6.25	7.81	11.34
4	0.711	1.064	1.923	3.357	5.39	7.78	9.49	13.28
5	1.145	1.610	2.675	4.351	6.63	9.24	11.07	15.09

Table 17.3

---**[1]**

7. A pure-breeding long-wing red-eyed fly and a pure-breeding short-wing white-eyed fly were crossed. All the F1 offspring were long-wing and red-eyed. When members of the F1 generation were crossed the F2 generation included 27 flies with long wings and white eyes.

Which of the options, **A** to **D**, shows the observed results that most closely match the expected results for the number of long-wing red-eyed flies and short-wing red-eyed flies?

A 92 long-wing red-eye and 31 short-wing red-eye
B 27 long-wing red-eye and 29 short-wing red-eye
C 86 long-wing red-eye and 11 short-wing red-eye
D 27 long-wing red-eye and 88 short-wing red-eye

Your answer ☐

[1]

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

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

(i) the allele for choroideremia

---

[1]

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

---

[1]

(iii) the genotype of a carrier of choroideremia.

---

[1]

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

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

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

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

the REP-1 protein is synthesised ---

[2]

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

---

[2]

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

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

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

---

[1]

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

---

[1]

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

---

[3]

9. The haploid chromosome number in the koala, *Phascolarctos cinereus*, is 8.

Independent assortment of chromosomes in meiosis contributes to genetic variation in the gametes of the koala.

How many genetically different versions of koala gamete would it be possible for one individual to produce if independent assortment were the only source of genetic variation?

A 64
B 128
C 256
D 512

Your answer [ ] **[1]**

10. The sweet pea plant has been used to study inheritance since the nineteenth century. The seeds of the sweet pea can vary in colour and shape.
The gene that controls colour has two alleles:

- **Y** is dominant and produces yellow seeds.
- **y** is recessive and produces green seeds.

The gene that controls shape has two alleles:

- **R** is dominant and produces round seeds.
- **r** is recessive and produces wrinkled seeds.

In the nineteenth century, Gregor Mendel crossed a pea plant that was heterozygous for both seed colour and shape with a pea plant that had green and wrinkled seeds.

(i) List the gametes that would be produced by a sweet pea plant that was heterozygous for both seed colour and shape.

---

[1]

(ii) List the genotypes of the offspring that were produced from Mendel's cross and state the corresponding phenotypes.

genotypes

---

phenotypes

---

[2]

11. This question is about genetic control and selective breeding.

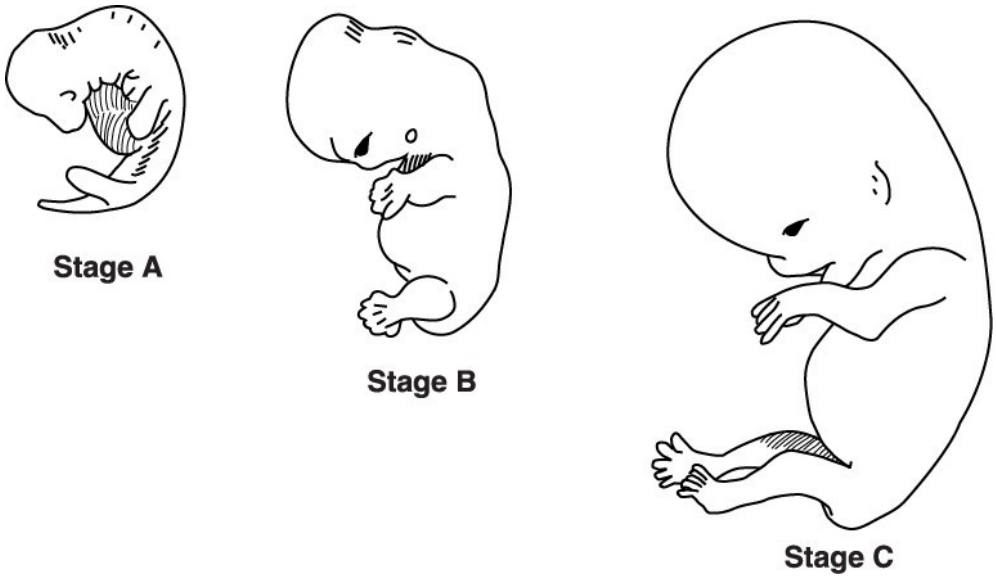
Genetic control is also achieved by genes switching on and off. There is a highly conserved set of genes called homeobox genes that control the development of body plans.

(i) Name **all** the **kingdoms** of living organisms that use homeobox genes to control the development of body plans.

---

[1]

This figure shows a human embryo during the second month of development.



(ii) How can the control of development by homeobox genes be seen in **Stage A** in the figure?

---

[1]

(iii) Describe how the process of apoptosis can be seen between **Stages A** and **B**, and between **Stages B** and **C**, in the figure.

Stages A to **B** ---

Stages B to C _____

[2]

12. The inheritance of different alleles in fruit flies, *Drosophila* spp., has been studied extensively in the laboratory.

Two genes that affect the appearance of *Drosophila* are:

R / r	red / pink eyes
Y / y	yellow / ebony body

Flies known to be heterozygous at both of these loci were crossed with homozygous pink-eyed ebony flies.

Based on the hypothesis that the two genes assort independently, the offspring expected from this cross would be four different phenotypes in a ratio of 1:1:1:1.
The results obtained, however, are shown in Table 4.2.

Phenotype	Expected number	Observed number
Red eye, yellow body	360	6
Pink eye, yellow body	360	701
Red eye, ebony body	360	729
Pink eye, ebony body	360	4

Table 4.2

The chi-squared (χ^2) test can be used to assess whether the results in Table 4.2 are significantly different from the expected results.

The equation for working out the value of χ^2 is given below.

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

where $\sum$ = 'sum of …'
O = observed value
E = expected value

(i) Calculate the value of χ^2 **to the nearest whole number** for the genetic cross results shown in Table 4.2.

Complete the table below and determine the value of χ^2.

Phenotype of fly	O – E	$(O - E)^2$	$\frac{(O - E)^2}{E}$
Red eye, yellow body	–354	125316	348
Pink eye, yellow body	341	116281	323
Red eye, ebony body			
Pink eye, ebony body			

χ^2 = _____

[3]

(ii) Statistical tables show that, for this data set, if χ^2 has a value of 11.35, the observed results would only be produced by chance in 1% of trials.

Use this information and the value for χ^2 that you have calculated in **(i)** to explain whether the original hypothesis should be accepted or rejected.

_____

_____

_____ **[1]**

(iii) The difference in the observed numbers from the cross compared with the expected numbers has **not** occurred by chance. Suggest a genetic explanation for this difference.

_____

_____

_____

_____

_____

_____

_____

_____ **[3]**

13. Hox genes code for transcription factors and control the development of the body plan. Fig. 16.2 shows a congenital deformity caused by failure of the control mechanism.

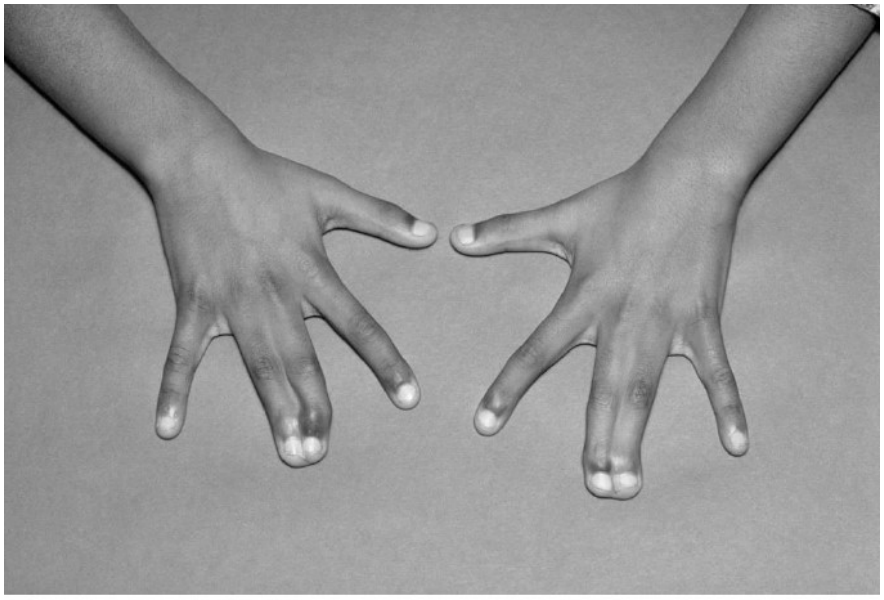


Fig. 16.2

Explain how failure of the control mechanism during development could lead to such a deformity.

---

[3]

14. The sweet pea plant has been used to study inheritance since the nineteenth century. The seeds of the sweet pea can vary in colour and shape.
The gene that controls colour has two alleles:

- **Y** is dominant and produces yellow seeds.
- **y** is recessive and produces green seeds.

The gene that controls shape has two alleles:

- **R** is dominant and produces round seeds.
- **r** is recessive and produces wrinkled seeds.

When Mendel crossed two pea plants that were heterozygous for both seed colour and shape, the ratio of phenotypes in the offspring was:

- 9 yellow round
- 3 green round
- 3 yellow wrinkled
- 1 green wrinkled.

Some students tried to recreate this investigation using a modern variety of plant that showed the same phenotypic variation in seed colour and shape.

The students crossed two of the modern plants that were heterozygous for both seed colour and shape. The results of this cross were:

- 58 yellow and round
- 31 green and round
- 21 yellow and wrinkled
- 2 green and wrinkled

The students used the chi-squared test to compare their data to the expected 9 : 3 : 3 : 1 ratio.

(i) Use the chi-squared formula $\chi^2 = \sum \frac{(O-E)^2}{E}$ to calculate the x^2 value for these data.
You may use the table below for working out.

χ^2=---------- [3]

Table 17 shows a x^2 probability table.

Degrees of freedom	Probability (p)					
	0.95	0.90	0.10	0.05	0.025	0.01
1	0.00	0.02	2.71	3.84	5.02	6.64
2	0.10	0.21	4.61	5.99	7.38	9.21
3	0.35	0.58	6.25	7.82	9.35	11.34
4	0.71	1.06	7.78	9.49	11.14	13.28
5	1.15	1.61	9.24	11.07	12.83	15.09
6	1.64	2.20	10.64	12.59	14.45	16.81
7	2.17	2.83	12.02	14.07	16.01	18.48

Table 17

(ii) After analysing the results, the students stated that the inheritance of the seed colour and shape in their investigation was different from that in Mendel's investigation.

Using Table 17, discuss whether the results of the investigation and the chi-squared test support the students' statement.

---

[3]

(iii) A ratio that is different from the expected 9 : 3 : 3 : 1, in a cross such as this, can be the result of epistasis.

Suggest and explain one reason, **other** than epistasis, why the phenotype ratio might not be 9 : 3 : 3 : 1.

Suggestion

Explanation

---

[3]

15. This question looks at two ways of using mathematical concepts in Biology.

When a new road system was constructed, it split a population of a rare snail species into three smaller populations, **A**, **B** and **C**. As a result, each of these populations became reproductively isolated.

The Hardy-Weinberg principle was used to calculate the relative frequencies, p and q, of a dominant and a recessive allele in each population.

Table 4.1 shows the values of p and q, and the estimated sizes of these three populations.

Snail population	Estimated population size	Immediately after road building		10 years after road building	
		p (frequency of dominant allele)	q (frequency of recessive allele)	p (frequency of dominant allele)	q (frequency of recessive allele)
A	1000	0.50	0.50	0.52	0.48
B	100	0.49	0.51	0.63	0.37
C	10	0.40	0.60	0.20	0.80

Table 4.1

(i) Name the type of isolating mechanism that prevents interbreeding between these three snail populations.

---------- **[1]**

(ii) The habitat of these snail populations did not change over the ten years.

State the term used to describe the **random** changes in allele frequency in a small population.

---------- **[1]**

(iii) Explain which of the populations, **A**, **B** or **C**, experienced most genetic change.

---------- **[2]**

16(a) In pigeons, the male bird is homogametic (XX) and the female bird is heterogametic (XY).

.

Feather colour in pigeons is a sex-linked characteristic that is affected by one gene which has three alleles.

In female birds allele C^r produces ash red feathers, C^{br} produces brown feathers and C^{bl} produces blue feathers.

C^r is dominant to C^{br}, which is dominant to C^{bl}.

A pigeon breeder crossed an ash red male with a brown female. The two eggs hatched to produce one brown male and one blue female.

Use a genetic diagram to explain these results.

	Male	Female
Parental genotypes:		
Gametes:		
F1 generation genotype:		
phenotype:		

[4]

(b). Pigeons can live for 15 years in captivity. They are kept in small mixed flocks but tend to be monogamous (have one partner). Each season the female produces two eggs.

A student used a genetic diagram to show the breeder that over a number of generations the following ratio of offspring could be expected from the breeding pair.

1 ash red male : 1 brown male : 1 ash red female : 1 blue female

The breeder decided to test this prediction.

Over a number of breeding seasons records were kept of the offspring produced by the same pair of birds. Table 17.1 shows the results recorded by the breeder.

Year	Males		Females	
	Ash red	Brown	Ash red	Blue
1		1		1
2	1		1	
3		1		1
5**	1			1
6	1	1		
7	2			
8		1	1	
9		1		1
10	1			1
11	1		1	
total	7	5	3	5

** in year 4 there were two brown female chicks

Table 17.1

The chi-squared test can be used to assess the probability of achieving these observed results. The value of chi-squared is given by the formula:

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

(i) Use Table 17.2 to calculate the value of chi-squared using the ratio predicted by the student as the expected results.

	Ash red male	Brown male	Ash red female	Blue female	Total
O	7	5	3	5	20
E	5	5	5	5	20
$(O–E)^2$					
$(O–E)^2 / E$					

Table 17.2

X^2 =_____

[2]

(ii) The critical value of chi-squared for three degrees of freedom at a probability of 0.05 is 7.81.

What can you conclude about the observed results collected by the breeder in Table 17.1?

_____[1]

(iii) Explain why the observed results did not exactly match the predicted results.

_____

_____[1]

(c). In year 4 the breeder noticed that the two chicks were brown feathered females. The student had not predicted that brown feathered females would be produced. The value of E for this category would be zero. Therefore the breeder had left this category out of the results table.

(i) What effect would adding this unexpected result into the results table have on the value of chi-squared?

---

[1]

(ii) Assuming that the student had made an accurate prediction about the ratio of offspring, what might the breeder have concluded about the parents of the chicks in year 4?

---

[1]

(iii) Explain how you have reached this conclusion.

---

[2]

17. The Hardy-Weinberg principle, represented by the equations below, can be used to estimate the frequency of alleles in a population.

$$p^2 + 2pq + q^2 = 1$$

$$p + q = 1$$

Albino rabbits have white fur as these individuals are unable to produce the pigment melanin. The ability to produce melanin is controlled by a gene with a dominant allele (B), resulting in brown fur, and a recessive allele (b), resulting in an albino.

Of the 60 rabbits in a pet shop, 45 are brown.

(i) A student decided to use the Hardy-Weinberg principle to estimate the frequencies of the alleles in this group of rabbits.

Using the Hardy-Weinberg equations, calculate the frequency of the dominant allele in this group.

Show your working.

Frequency of the dominant allele =_____

[3]

(ii) Give **two** reasons why it was not appropriate to use the Hardy-Weinberg principle to estimate the frequencies of alleles in this group of rabbits in the pet shop.

1

_____

_____

2

_____

_____

_____

[2]

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

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

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

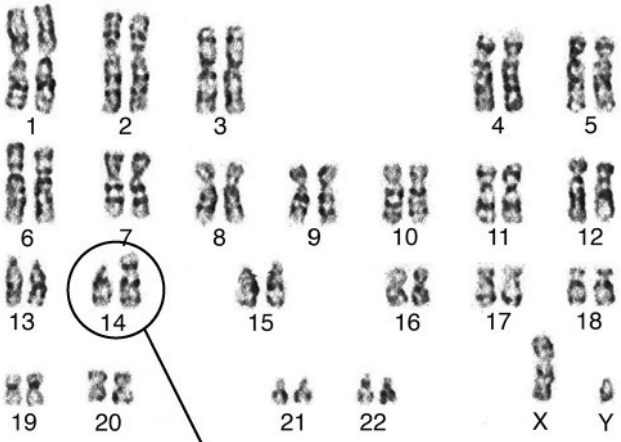


Fig. 2.1a

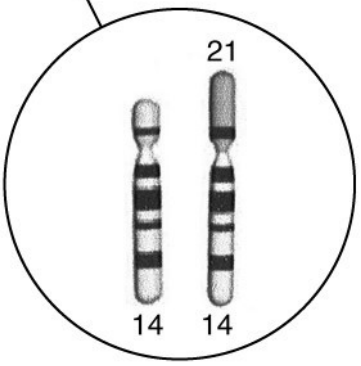


Fig. 2.1b

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

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

[7]

19. One subspecies of tiger is the Bengal tiger. One in 10 000 Bengal tiger births results in a white Bengal tiger.

White Bengal tigers (as shown below and in Fig. 3.1b **on the insert**) have black stripes but lack orange fur.

Fig. 3.1b

The allele that causes white fur is recessive and is a result of a mutation to a gene called SLC45A2.

According to the Hardy-Weinberg principle, the following equations can be used to estimate allele frequency within a population:

$$p^2 + 2pq + q^2 = 1$$

$$p + q = 1$$

Use the Hardy-Weinberg equations to calculate the percentage of Bengal tigers that are heterozygous for the SLC45A2 gene.

Give your answer to **one** significant figure.

Show your working.

Answer: _____ % **[3]**

20(a) The cheetah, *Acinonyx jubatus*, is a member of the cat family, Felidae.

.

Cheetahs display less intraspecific variation than other members of the family Felidae.

Fig. 20.1 shows the mean body length of a population of cheetahs from southern Africa.

The error bars on Fig. 20.1 show the standard deviation of mean body length.

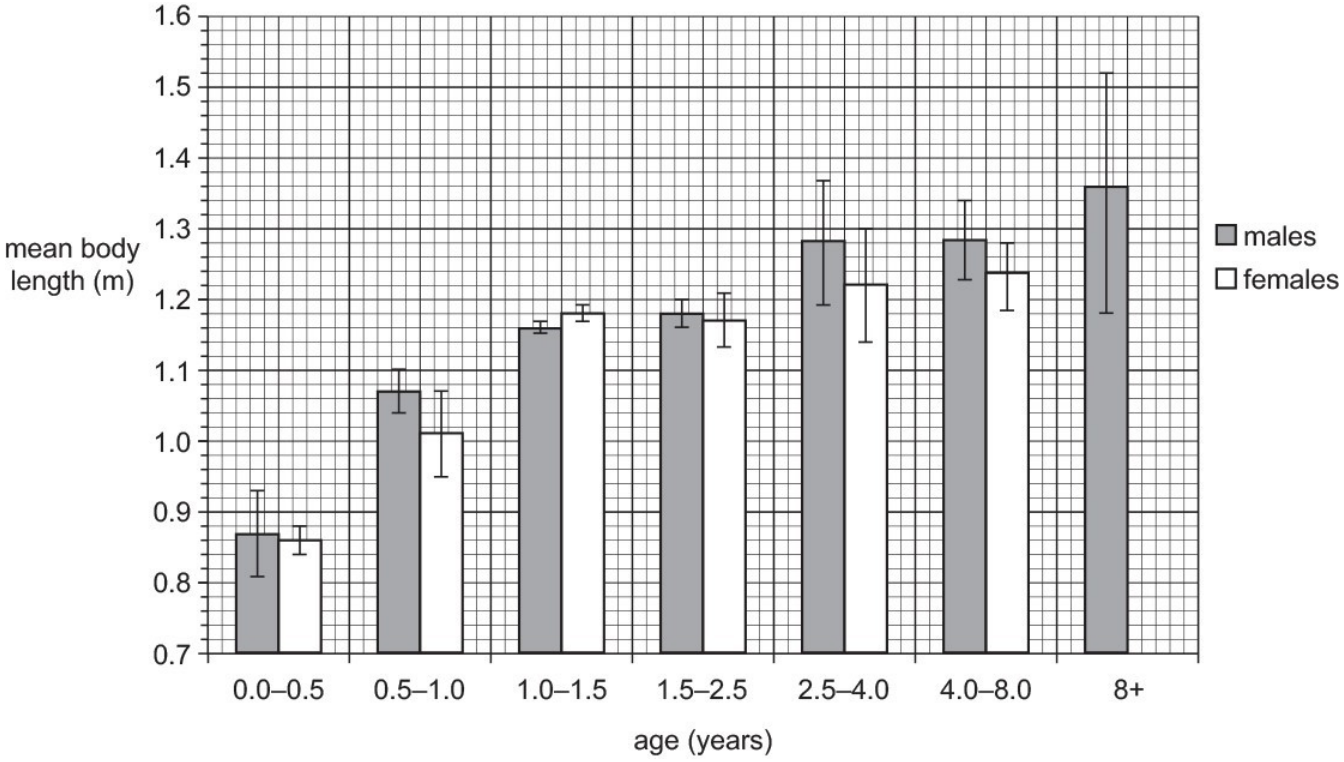


Fig. 20.1

(i) At between 2.5 and 4 years old, the mean length of female cheetahs is less than that of males.

Calculate how much shorter than males female cheetahs are.

Show your working. Express your answer as a percentage to **two significant figures.**

Answer ..... % **[2]**

(ii) Using only Fig. 20.1 and your answer to (i), what can be concluded about the **significance** of the difference

between the length of male and female cheetahs aged between 2.5 and 4 years?

Explain your answer.

---

[2]

(iii) A student looked at Fig. 20.1 and wrote:

"The longest male cheetah that was measured was 1.52 m long".

Explain whether the information in Fig. 20.1 supports the student's answer.

---

[1]

(iv) State the likely causes of variation in body length in cheetahs.

---

[2]

(b). Madagascar is a large island off the coast of Africa that once formed part of the mainland.

The fossa, *Cryptoprocta ferox* is the top predator on Madagascar.

The fossa shares many physical similarities with cats but it is not a member of the family Felidae. It is related to the mongoose.

The mongoose is a much smaller mammal that lives on the African mainland.

Fig. 20.2 shows a fossa and a mongoose.

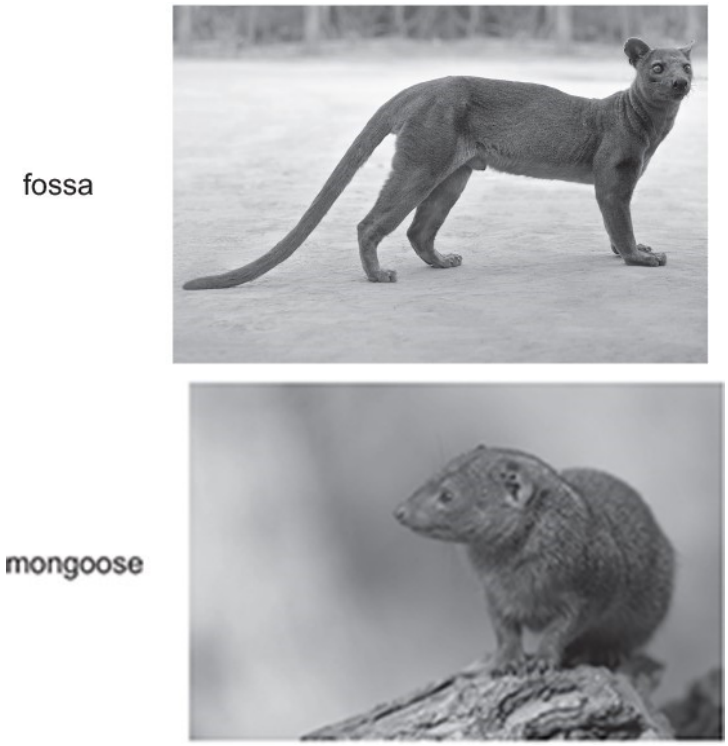


Fig. 20.2

(i) The mongoose is a smaller mammal and also has proportionally longer fur. State **one** other difference, **visible in Fig. 20.2**, between a fossa and a mongoose.

---

[1]

(ii) When the island of Madagascar became separated from the African continent, there were no members of the cat family, Felidae, on the island.
Outline how a fossa could have evolved from a much smaller, mongoose-like ancestor.

[4]

(iii) Islands, such as Madagascar, often have species that are different from those on the nearest land mass because they are reproductively isolated.
State **three** other conditions that must be present in order for speciation to occur.

1

2

3

[3]

END OF QUESTION PAPER