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.

.....

.....

.....

.....

.....

.....

.....

.....

.....

..... [4]

(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.

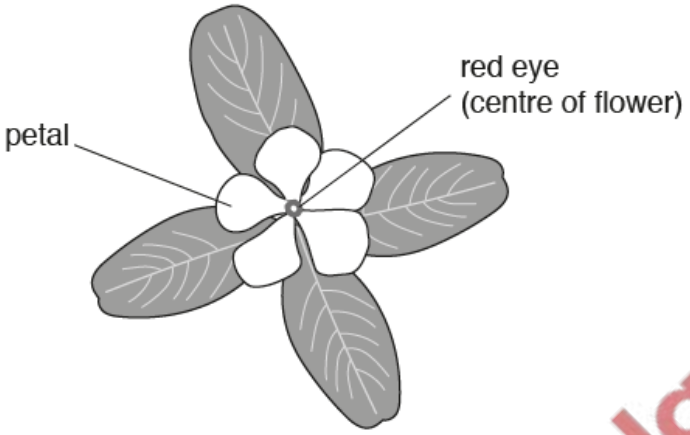

Fig. 4.1

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]

(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.

.....

.....

.....

.....

.....

.....

.....

.....

.....

..... [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.

.....

.....

..... [1]

[Total: 15]

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.

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.

.....

.....

.....

.....

.....

.....

.....

.....

.....

..... [4]

(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.

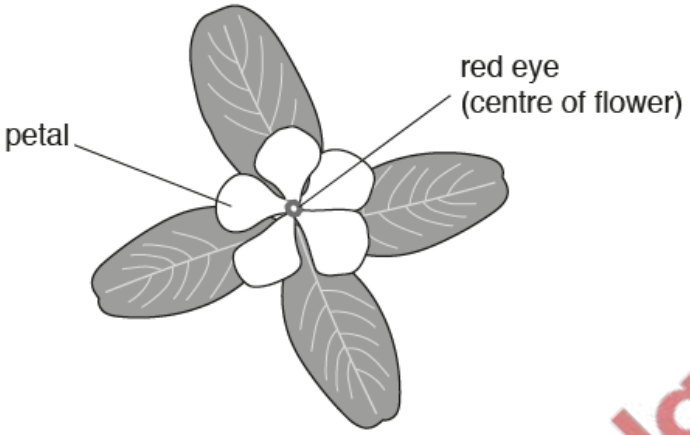

Fig. 4.1

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]

(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.

.....

.....

.....

.....

.....

.....

.....

.....

.....

..... [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.

.....

.....

..... [1]

[Total: 15]

3. Nov/2019/Paper_43/No.2

(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.

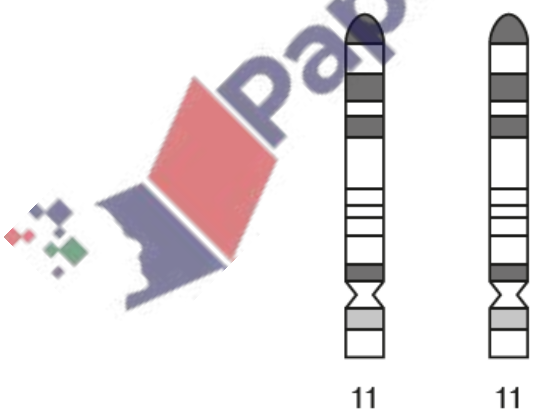

Fig. 2.2

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

.....

.....

.....

.....

.....

..... [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.

.....

.....

.....

..... [2]

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

.....

.....

.....

.....

..... [2]

(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.

.....

.....

.....

.....

.....

..... [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]

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.

.....

.....

.....

.....

..... [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.

.....

.....

.....

.....

..... [2]

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

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

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.

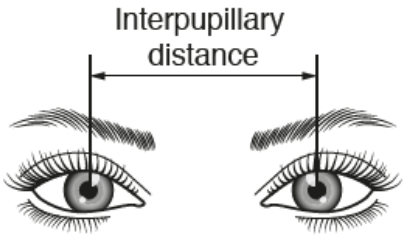

Fig. 2.1

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.

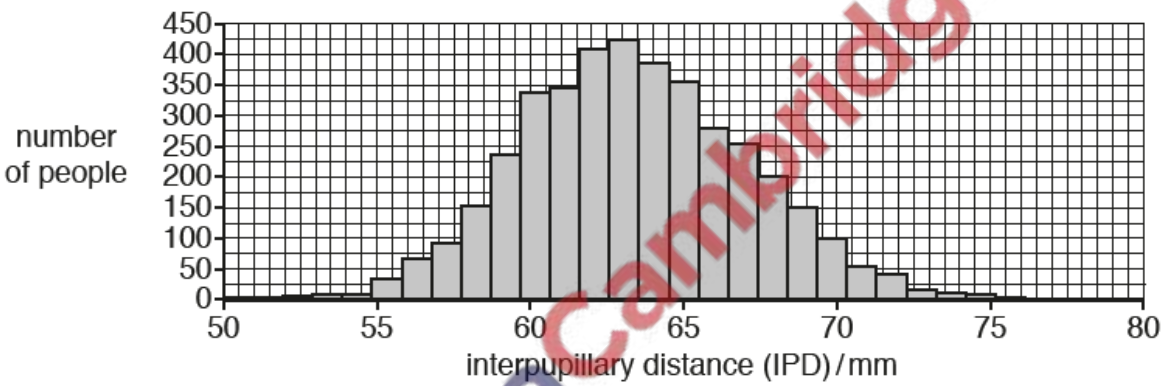

Fig. 2.2

(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.

.....

.....

.....

.....

.....

..... [3]

(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.

.....

.....

.....

.....

.....

..... [3]

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

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

..... [5]

(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.

.....

.....

.....

.....

.....

.....

.....

..... [3]

[Total: 15]

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

.....

.....

parental genotypes

gametes

offspring genotypes

ratio of offspring phenotypes

[4]

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

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

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]

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

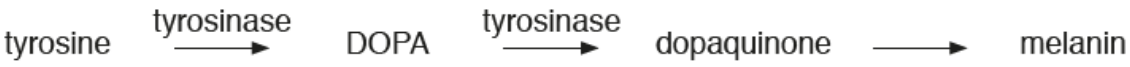

Fig. 4.1

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.

.....

.....

.....

..... [2]

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

.....

.....

.....

.....

.....

.....

.....

..... [4]

(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.

.....

.....

.....

..... [2]

[Total: 12]

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.

.....

.....

..... [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.

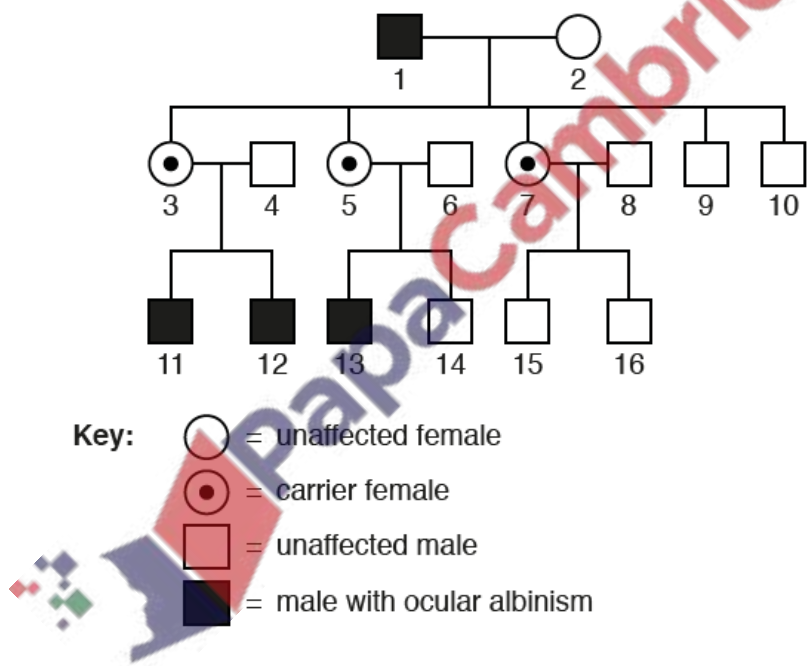

Fig. 2.1

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

.....

.....

.....

.....

.....

.....

..... [3]

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

key to symbols

.....

.....

parental genotypes

gametes

offspring genotypes

offspring phenotypes

[4]

(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.

.....

.....

.....

.....

..... [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 .....

.....

[2]

[Total: 13]

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.

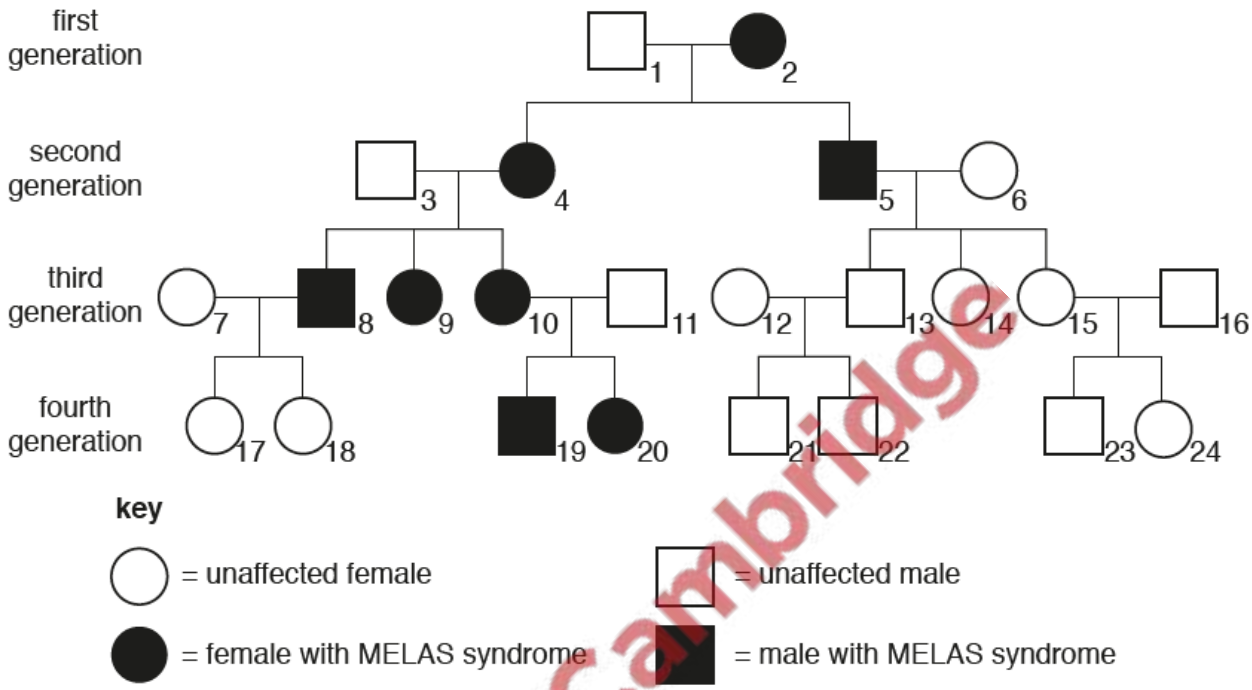

Fig. 6.1

(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.

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

..... [4]

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

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