Inheritance – 2019 Biology A2 9700

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.

Meiosis produce 4 cells in both gametogenesis of plants and animals. Meiosis produce haploid cells in both plants and animals. Cells produced in meiosis are genetically different. However, a sperm cell is a gamete whereas pollen grains not a gamete. A sperm cell has one nucleas while a pollen grain has two haploid nuclei.

[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.

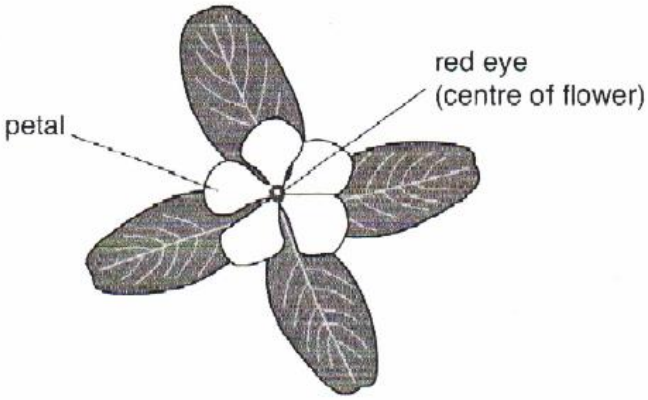

Fig. 4.1

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 ..... Pink pale petals, red eye .....

Rr Dd Pp ..... dark pink petals, red eye .....

rr Dd Pp ..... white petals, no red eye .....

RR dd pp ..... White petals, red eye ..... [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.

A change in base sequence will cause a change in every triplet ^codon from the site of deletion. This will change the codes for the amino acids in proteins. Then transcription produce mRNA with a different code. So, the protein produced will have 3 dimension shape. A stop codon may form too early leading to formation of a shorter polypeptide. A protein with a changed tertiary structure will produce an enzyme that cannot bind to substrate. So biosynthetic pathway does not function.

[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.

It is a promoter region

[1]

[Total: 15]

2. 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.

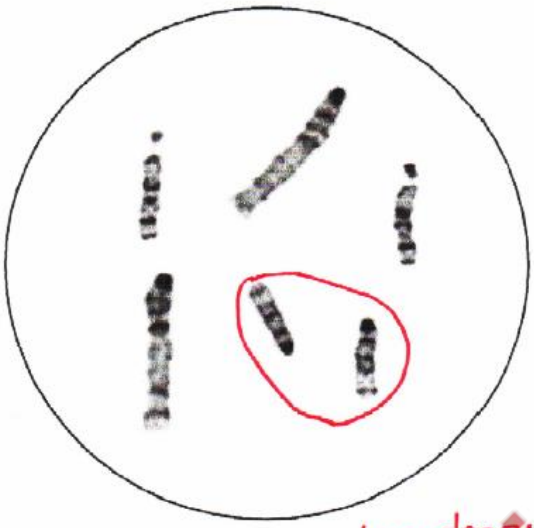

Fig. 2.1

homologous chromosomes have similar shape and lengths

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.

Homologous chromosomes have same length and size. When stained, they show same banding pattern. This implies they have same gene loci. The homologous chromosomes pair up to form a bivalent.

[3]

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

20 [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.

Meiosis halves number of chromosome in diploid cells to form haploid cell. This prevents chromosomes from doubling at fertilisation

[2]

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

Gametes fuse by random chance. So, a gamete with a recessive gene can be fertilised so that a homozygous zygote is formed.

[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.

Mutation may produce a code for STOP codon. The stop codon will stop the translation process so that no amino acid is added to a growing polypeptide chain.

[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

Aa x Aa

gametes

A a A a

offspring genotypes

AA Aa Aa aa

offspring phenotypes

Normal normal normal woody

Normal Normal Normal woody

3 Normal : 1 woody

[3]

[Total: 14]

3. 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.

Le

..... [1]

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

The dominant allele codes for an enzyme that converts gibberellin into 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.

Growth genes cause elongation and enlargement of cells. This is due to mitosis, a cell division that causes growth.

..... [2]

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

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

Huntington's disease is caused by a dominant mutation in a gene located in chromosome 4. The gene codes for Huntington protein. Mutation produce a code for abnormal Huntington protein. The code for normal Huntington protein has 10-35 CAG repeats.

However, abnormal protein is coded for by extra CAG. Larger repeats of CAG cause an earlier onset of Huntington's disease. The usual onset of the disease is normally at 28, middle age but before 65.

The onset of the disease is from 1 year old in babies. if there are numerous repeats of CAG. This leads to production of extra glutamine. So the protein formed misfolds leading to Huntington's disease. So the abnormal protein cause death of neurones.

Death of neurones cause uninhibited motor control. So there is involuntary movements called chorea. The disease Mood changes and loss of cognitive behaviour. The GABA producing neurons get lost.

5. 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.

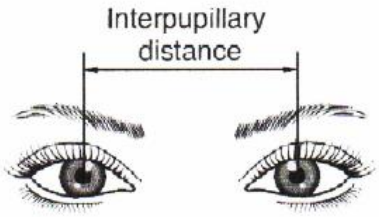

Fig. 2.1

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.

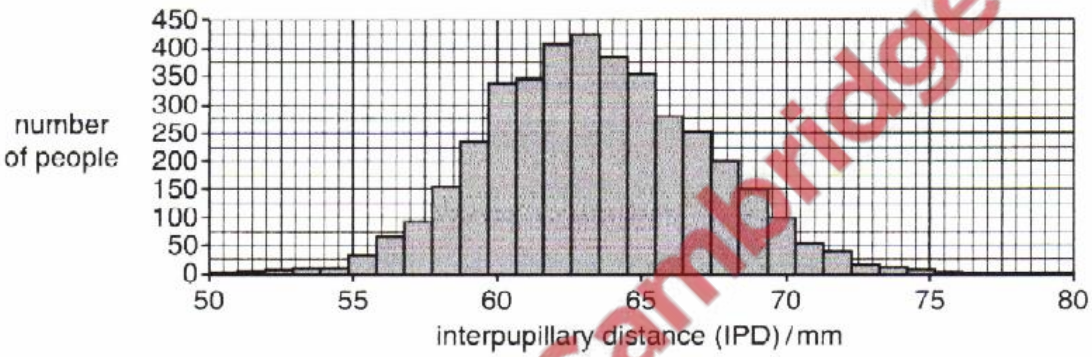

Fig. 2.2

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

Continous - it has intermediate values and is represented as normal distribution [1]

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

Interactions between genetics and the environment is the basis of continous variation. Different alleles at a single locus have a small effect on the phenotype and these added together cause additive effect. Environmental factors affect gene expression. Many genes have a combined effect on phenotype. [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.

PAX3 gene is a regulatory gene because it codes for transcription factor. PAX3 protein bind to DNA. The binding site for TF is a promoter which stimulates RNA polymerase binding

[3]

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

Get total mRNA from embryonic cell expressing PAX3. Then make cDNA from the mRNA. A fluorescent dye is then added to tag cDNA. The microarray has specific DNA spots from known genes. The cDNA is added to the spots so that it binds to the complimentary base pairs. This is called hybridisation. Fluorescent spots form in regions of hybridisation to show gene expression. The excess cDNA is washed off after hybridisation. Intensity of fluorescence gives the quantitative measure of gene expression.

[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.

A small genetic difference (PAX3) gives big phenotypic effects. This is because PAX 3 controls many other genes. The level of expression of other genes in chimpanzee affects the proteins they produce. Higher expression of PAX3 makes the chimpanzee's eyes to be closer together. This decreases its face width. [3]

[Total: 15]

6. 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

Normal allele = E

EOPD allele = e

parental genotypes

Ee x Ee

gametes

E e E e

offspring genotypes

EE Ee Ee ee

3 normal EOPD

ratio of offspring phenotypes

3 Normal : 1 EOPD

[4]

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

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

9 B.

The FSH is secreted by anterior pituitary gland. It stimulates growth cells within the follicles at the ovary. Eggs develope and mature within the follicles in the ovary.

Oestrogen is secreted by the Graafian follicle. It stimulates repair of the endometrium after menstruation. Secretion of oestrogen inhibit further release of FSH. Oestrogen levels increase in blood by day 14 after menstruation.

Oestrogen stimulates release of Luteinizing Hormone which in turn stimulates ovulation. Ovulation is the release of oocyte. Luteinizing hormone stimulates developement of Corpus luteum. Corpus luteum secretes progesterone.

Progesterone increase build up of the endometrium. Secretion of progesterone inhibits secretion of LH and FSH. When Corpus luteum degenerates, concentration of progesterone falls. So the endometrium breaks down.

8. June/2019/Paper_42/No.4

Mexican spadefoot toads, *Spea multiplicata*, live on land but return to ponds to breed. Eggs are laid in water and hatch into tadpoles, which feed in ponds before developing into adults.

The tadpoles can be classified into two main types: omnivore-type tadpoles and carnivore-type tadpoles. Differences between the phenotypes of these two types of tadpole are related to their different feeding behaviours.

- Omnivore-type tadpoles feed on tiny pieces of detritus (dead material from plants and animals) and algae (microscopic photosynthetic organisms) at the bottom of ponds. These tadpoles grow slowly.
- Carnivore-type tadpoles feed on small animals in the water, such as fairy shrimp and small omnivore tadpoles. These tadpoles grow quickly.

Fig. 4.1 shows two tadpoles of the same age, one of each type. A fairy shrimp is also shown. All three organisms are at the same distance from the camera.

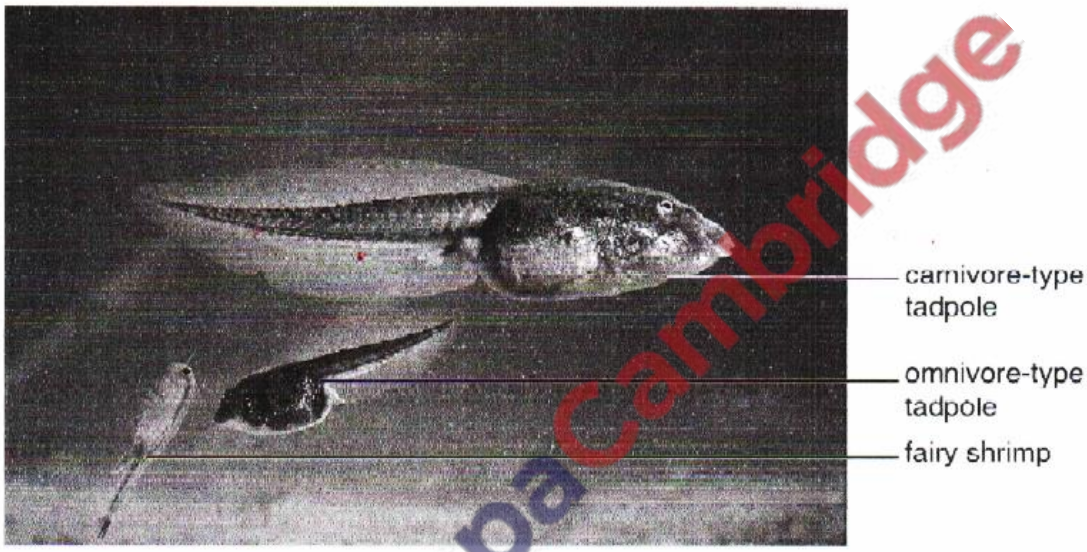

Fig. 4.1

Between these two main types of tadpole there is a continuous range of tadpoles with intermediate body phenotypes and feeding behaviours. Continous variation

For any individual tadpole, regardless of age, it is possible to calculate a phenotype score depending on the features of the tadpole. A tadpole with a phenotype score close to 3 is a typical omnivore type and a tadpole with a phenotype score close to 7 is a typical carnivore type.

The phenotype scores were determined for a large number of tadpoles sampled from two ponds. The availability of detritus and algae was high for one pond and low for the other pond. All other conditions in the two ponds were similar.

For both ponds, phenotype scores were determined shortly after the tadpoles had hatched from eggs and ten days later.

(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.

Missense results in a different amino acid. A change in the amino acid in a polypeptide chain results in a different protein. A stop codon may form too early leading to truncated translation process. [2]

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

Insertion of TYR changes the frameshift of the gene sequence in the DNA. This alters the sequence of amino acids produced at the ribosomes. This affects the primary structure of the protein produced. So the enzyme tyrosinase may fold incorrectly. This may introduce a STOP codon, which leads to formation of shortened polypeptide. The tyrosinase may not have a non-functional active site. So tyrosinase will fail to convert to DOPA, then DOPA does not form dopaquinone. So melanin will not form. [4]

(c) Worldwide, 1 in 17000 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.

They are geographicaly isolated from other human populations. So they have inbreeding within their population. They have a small gene pool. Albinism has a selective advantage among them. [2]

[Total: 12]

9. 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.

Albinism is a condition characterised by lack of melanin. So the hair of an albino individual is white and the skin is pink and eyes very pale. [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.

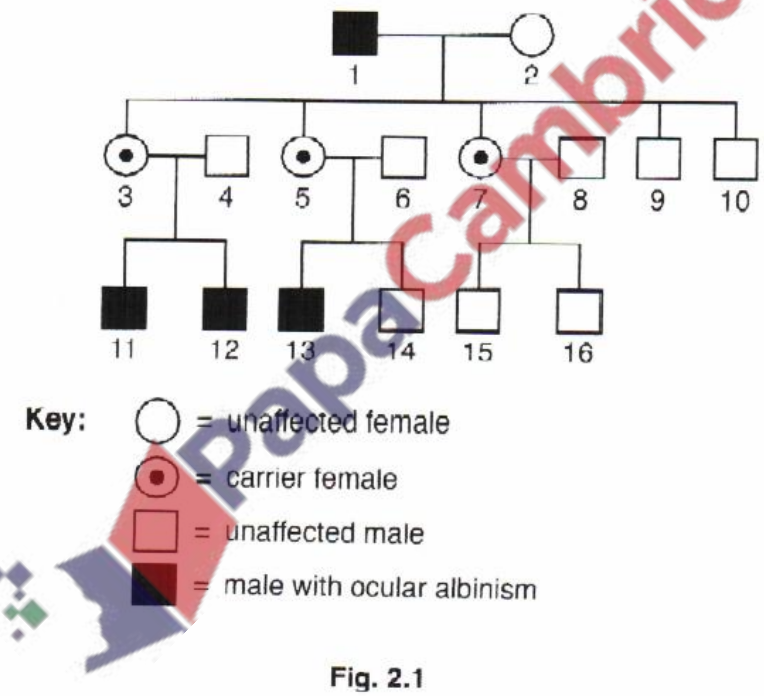

Fig. 2.1

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

More males have ocular albinism. The males with one copy of recessive allele have ocular albinism. The females with a copy of recessive allele are carriers. It is the mothers that pass on ocular albinism to their sons. This is because fathers pass Y chromosomes to their sons. [3]

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

key to symbols

A

a

parental genotypes

$X^aY \times X^AX^A$

gametes

$X^a \quad Y \quad X^A \quad X^A$

offspring genotypes

$X^AX^a \quad X^AY$

offspring phenotypes

Carrier female Normal male

[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.

This causes a change in every triplet from the site of deletion. The protein formed during translation at ribosome will have a different primary structure. So the binding site of the protein will be affected. A STOP codon may form too early leading to incomplete 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 The test helps the woman decide wether to have children or not.

against The test may be too expensive

[2]

[Total: 13]

10. 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.

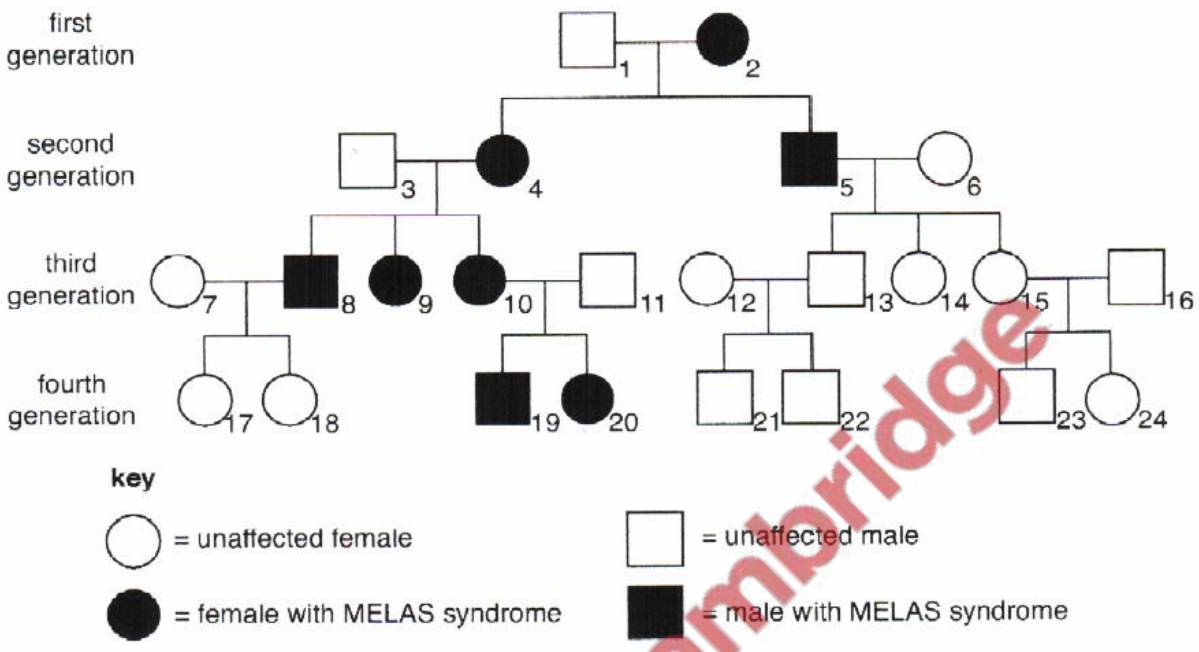

Fig. 6.1

(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.

MELAS syndrome is not inherited from affected males. All offspring of a female with MELAS syndrome also have MELAS syndrome. So it is not caused by gene mutation.
It is linked to X chromosome because more males are likely to be affected. This is true because males inherit only one X chromosome from the mother. Even so, more females are affected than males. Therefore, there are no heterozygous females that carry the genes for MELA syndrome. [4]

(b) Analysis of mtDNA can show how recently species have evolved from each other.

Describe the properties of mtDNA that make it suitable for the study of evolution.

In mitochondrial DNA, mutations occur at a constant rate. At the mitochondria, the DNA are not protected histone proteins. There are no enzymes at the mitochondria to repair DNA mutations. There are many copies of mitochondrial DNA per cell.

[3]

[Total: 7]

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

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

10B.

Meiosis shuffles the genetic material in a way that leads to formation of new allele combinations. This produce different versions of genes. This contributes to the diversity of traits in an organism.

Meiosis produces different alleles through genetic recombination. Homologous chromosomes pair up and exchange segments of DNA during the first meotic cell division (Prophase I). This makes the non sister chromatids to exchange genetic material at the chiasmata. This forms new combination of alleles

In metaphase I, the homologous chromosomes line up at the equator. They get pulled to appart randomly so sister chromatids are assorted independently. This causes chromosomal mutations.

During fertilisation, gametes are fertilized randomly. Then a diploid zygote is formed. At fertilization there is sharing of genetic information the genes in the gametes will affect the phenotype of the zygote.

Meosis produce genetic variation that can be selected by natural selection. This allows species to adapt to the changing conditions of the environment. This helps to drive process of evolution over time.