MUTATION

QUESTIONSHEET 1

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New variations may sometimes arise because of mutation. This may be gene mutation or chromosomal mutation.

(a) Briefly distinguish between gene and chromosomal mutations.

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.....

[3]

(b) Name two specific types of gene mutation and give two examples of gene mutation.

Type 1: .....

Type 2: .....

Examples: .....

.....

[4]

(c) What is Down's syndrome?

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[2]

(d) When mutations occur they usually cause discontinuous variation. However, continued gene mutation can lead to continuous variation. Explain how this is so.

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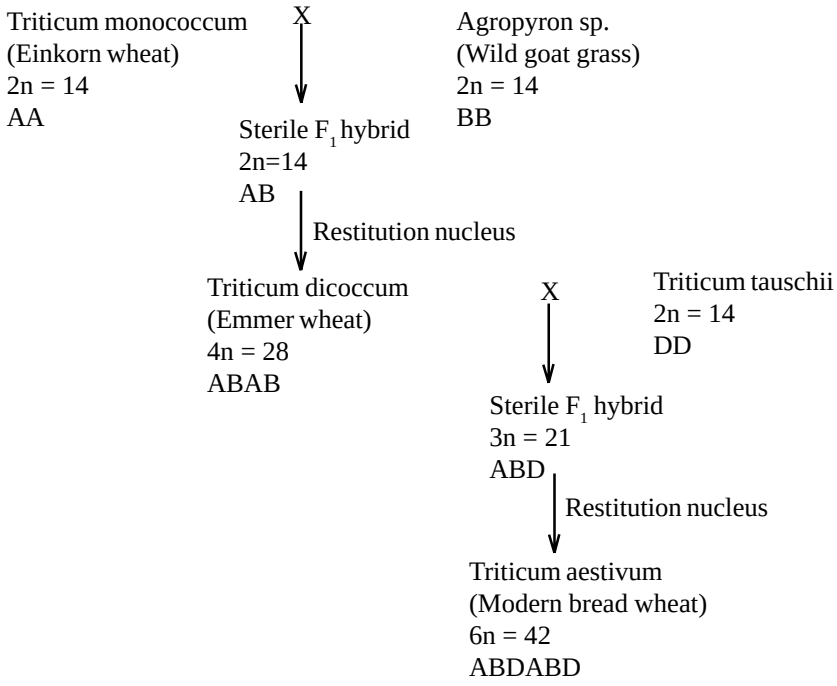
[2]

TOTAL / 11

Do not write in margin

The flow diagram below illustrates the evolution of modern Bread Wheat, Triticum aestivum which has occured during the last 10,000 years. Modern Bread Wheat has arisen by polyploidy occuring between ancient cultivated wheats and wild species.



(a) (i) What is meant by the term ‘polyploidy’?

.....

.....

[2]

(ii) The type of polyploidy illustrated above involves hybridisation. What name is given to this type of polyploidy?

.....

[1]

(iii) What do the letters A, B and D indicate?

.....

[1]

(iv) What is a ‘restitution nucleus’?

.....

.....

[2]

(Continued...)

Do not write in margin

(b) Unlike other forms of speciation which are gradual, polyploidy can produce new species within one generation. Explain why this is so.

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.....

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[3]

(c) (i) Polyploid plants have evolutionary advantage since they possess 'hybrid vigour'. What is meant by 'hybrid vigour'?

.....

.....

.....

[3]

(ii) Einkorn wheat was cultivated by Neolithic humans and Emmer wheat by ancient Iranian peoples. Suggest two advantages you think Modern bread wheat may have over the ancient breeds?

1: .....

2: .....

[2]

TOTAL / 14

MUTATION

QUESTIONSHEET 3

Suggest mechanisms to explain the following.

(a) The emergence and establishment of new strains of DDT-resistant mosquitoes.

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[4]

(b) The emergence of antibiotic strains of bacteria among many different bacterial species.

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[4]

(c) The sudden emergence of fertility in a sterile F_1 hybrid.

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[2]

TOTAL / 10

QUESTIONSHEET 4

(a) What is 'background radiation' and why is it present?

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.....

[2]

(b) Distinguish alpha, beta and gamma radiation and say which is least likely to cause mutation.

.....

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.....

[4]

(c) Name two chemical mutagens.

1: .....

2: .....

[2]

(d) (i) The damage caused by radiation to the genetic material is cumulative. What does this mean?

.....

.....

[1]

(ii) A sample of Drosophila (fruit flies) was irradiated with X-rays giving a dose of 5 Sieverts (a measure of radiation dose). The mutation rate of a specific gene was found to be increased by 2%. What would the expected percentage mutation rate be after a radiation dose of 12.5 Sieverts? Show your working.

Answer:..... [2]

(e) (i) List three types of gene mutation.

1: ..... 2: ..... 3: .....

[1]

(ii) List three types of chromosome mutation where chromosome structure is altered.

1: ..... 2: ..... 3: .....

[1]

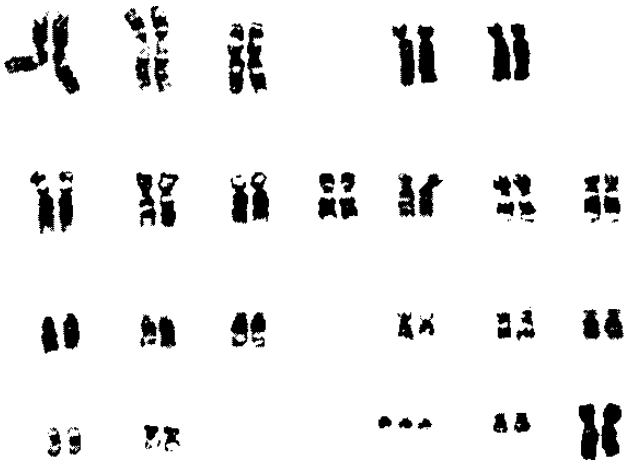
TOTAL / 13

MUTATION

QUESTIONSHEET 5

Do not write in margin

The picture below shows human chromosomes obtained by karyotyping.



(a) (i) Describe the process of karyotyping.

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.....

.....

.....

.....

[4]

(ii) In humans karyotyping is usually carried out after amniocentesis. What is amniocentesis?

.....

.....

.....

.....

[3]

(*Continued...*)

Do not write in margin

(b) (i) Does the karyotype above represent a full set of human chromosomes? Explain your answer.

.....

.....

.....

[2]

(ii) A mutation is shown in the above karyotype. Label it on the drawing, name it and explain how could it have happened.

Name: .....

How it happened: .....

.....

.....

.....

[4]

(iii) What was the sex of the individual from which the karyotype was taken? Explain your answer.

Sex: .....

Explanation: .....

.....

[2]

TOTAL / 15

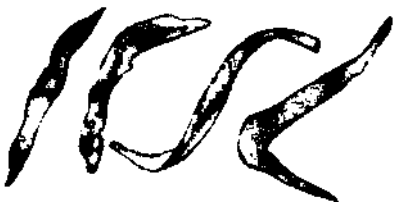
MUTATION

QUESTIONSHEET 6

Do not write in margin

The drawings below show the appearance of normal human red cells and red cells from an individual with sickle cell anaemia. The condition is caused by a mutant recessive gene.In the homozygous condition it causes sickle cell anaemia which has serious disabling effects on sufferers, while in the heterozygous condition it causes sickle cell trait which is harmless and gives resistance against infection by malarial parasites. The abnormal gene also changes the permeability of red cell membranes so that the red cells lose potassium ions.

normal red cells 

sickle cells 

(a) The mutation affects the amino acid sequence of the β-globin chain of haemoglobin. The normal amino acid sequence, together with the mRNA and DNA bases which code for the sequence are shown below.

amino acid sequence:	valine	/histidine	/leucine	/threonine	/proline	/glutamic acid	/glutamic acid
mRNA codons:	GUA	CAU	CUC	ACU	CCA	GAA	GAA
DNA bases:	CAT	GTA	GAG	TGA	GGT	C**T**T	CTT

(i) If the base marked with an arrow was changed to A (adenine) what would be the effect on the amino acid sequence of the globin? Explain your answer.

.....

.....

..... [3]

(ii) What will be the probable effect is this change in amino acid sequence on the structure of the haemoglobin? Explain your answer.

.....

.....

..... [2]

(iii) Name this type of gene mutation.

..... [1]

(b) The sickle cell haemoglobin is known as haemoglobin S. This type of haemoglobin is found mainly in individuals who live in the malarial belts of the world or in their descendents. Suggest and explain why this is so.

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.....

..... [4]

TOTAL / 10

MUTATION

QUESTIONSHEET 7

Cystic fibrosis is the most common recessive genetic disorder in the UK Caucasian population with an overall birth frequency of about 1 in 2500. It is characterised by severe respiratory problems and inadequate pancreatic function, caused by accumulation of sticky mucus.There is no cure at present but improved treatments can now increase the life expectancy to about 30 years, death usually being due to respiratory failure. Males with CF are usually sterile. Cystic fibrosis is due to a mutation in the gene encoding the cystic fibrosis transmembrane conductance regulator(CFTR). This protein regulates transport of chloride across cell membranes. The mutant gene is recessive and heterozygotes do not exhibit symptoms. The mutant gene occurs in about 4% of the population and usually arises by deletion of three adjacent nucleotides.

(a) (i) Explain how the deletion could occur and influence the effectiveness of the CFTR gene.

.....

.....

.....

.....

..... [4]

(ii) Name two other types of gene mutation.

1: ..... 2: ..... [2]

(b) (i) By means of a genetic diagram show the probability of two carrier parents producing a CF child.

Probability: ..... [5]

(ii) What would be the expected % incidence of cystic fibrosis in the population? Explain your answer.

.....

.....

.....

.....

..... [4]

(iii) Since cystic fibrosis is a lethal condition,why does its allele persist in the population?

..... [1]

(c) Suggest two treatments for cystic fibrosis.

1: .....

2: ..... [2]

TOTAL / 18

MUTATION

QUESTIONSHEET 8

Do not write in margin

About 80% of the world's coffee is obtained from Coffea arabica. The genus Coffea contains about 100 species of small evergreen seed producing trees, with a basic diploid chromosome number of 2n = 22. These species are generally outbreeding. The seeds are the coffee beans that can be processed and ground for use. Coffea arabica, however, is a tetraploid with 2n = 44 and it is generally inbreeding, a habit which, in this case, provides considerable advantages.

(a) (i) Explain how the tetraploid condition could have arisen in Coffea arabica.

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..... [4]

(ii) Coffea arabica is genetically isolated from its closely related species and ancestral species, with which it cannot successfully interbreed. Explain why this is so.

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..... [3]

(b) Suggest four reasons why the inbreeding habit of Coffea arabica has been advantageous.

1: .....

2: .....

3: .....

4: .....

[4]

TOTAL / 11

MUTATION

QUESTIONSHEET 9

Do not write in margin

Read through the following passage concerning grapefruit (Citrus paradisii) and then answer the questions below.

Grape fruit originated on the island of Barbados in the middle of the 1700s. It arose as a result of hybridisation between the orange (Citrus sinensis) and the pummelo (Citrus grandis). Grape fruit have apomictic seeds, meaning that the seeds develop without being fertilised. Red 'ruby' grapefruit arose in Texas in the 1920s as a result of mutation in a white-fleshed variety.

(a) Describe what is meant by the term 'hybridisation' in the passage above.

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.....

.....

..... [3]

(b) Suggest why the grapefruit reproduces by apomictic seeds.

.....

.....

..... [2]

(c) What will the consequences be to the grapefruit of having apomictic seeds?

.....

.....

..... [2]

(d) What type of mutation would have produced the 'ruby' variety?

.....

..... [2]

TOTAL / 9

MUTATION

QUESTIONSHEET 10

The nectarine arose as a single gene mutation in a peach which caused the development of a smooth skin instead of the 'fuzzy' skin of a peach.

(a) Explain what is meant by the term 'single gene mutation'.

.....

.....

.....

..... [3]

(b) (i) Name two conditions in humans that are caused by single gene mutations.

1. .....

2. ..... [2]

(ii) Describe the genetic basis of Down's syndrome in humans.

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.....

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..... [3]

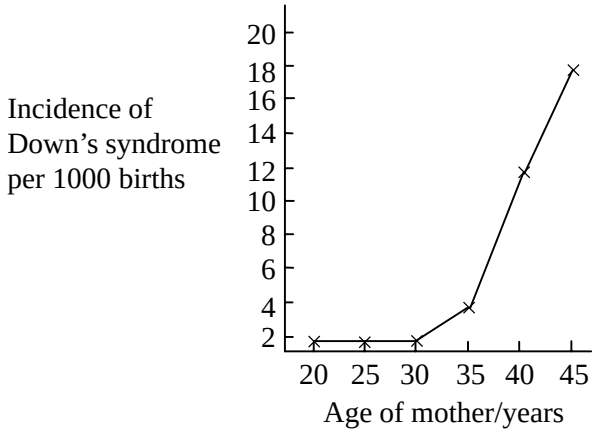
TOTAL / 8

MUTATION

QUESTIONSHEET 11

Do not write in margin

The graph below shows the effect of mother's age on the incidence of children born with Down's syndrome.



(a) (i) Describe the trend in the incidence of Down's syndrome shown in the graph.

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..... [3]

(ii) Suggest a reason for the change in the incidence of Down's syndrome.

.....

..... [1]

(iii) Down's syndrome is also known as trisomy 21, Edward's syndrome is trisomy 18 and Patau's syndrome is trisomy 13. Describe how these trisomies may arise.

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..... [3]

(b) Women over 40 years of age are advised to have the chromosomes of the fetus examined. Suggest how this could be done.

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.....

.....

..... [3]

TOTAL / 10

MUTATION

QUESTIONSHEET 12

Carefully study chromosomes A and B. Chromosome B differs from chromosome A due to a chromosomal mutation.

A

B

(a) (i) Name the type of chromosomal mutation that has occured.

..... [1]

(ii) Draw an arrow (◄——►) under chromosome B to show the mutated region. [1]

(b) Chromosome B is from a human and chromosome A is the equivalent chromosome from a chimpanzee.

(i) How do the chromosomes show that humans and chimpanzees are closely related genetically, almost certainly sharing some common ancestry in their evolutionary history?

.....

.....

.....

..... [3]

(ii) Altogether there are nine similar mutations altering the genotypes of man and chimpanzees. At what stage in the evolutionary history of these species did the mutations probably occur?

..... [1]

TOTAL / 6