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| *Human Perspectives ATAR Units 3 & 4* |

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Worksheet 9.1 Mutations

Answers

Use Chapter 9 of *Human Perspectives ATAR Units 3 & 4* to complete this flow chart summary of the topic of mutations.

*Answer:* An agent which is known to increase the rate that mutations occur. For example: mustard gas, formaldehyde, sulfur dioxide, antibiotics, ionising radiation, nuclear explosions.

*Answer:* An organism with a characteristic resulting from a mutation

Mutagens

Gene mutations

Point

Lethal recessive

Germ line

Somatic

Mutations

Chromosomal mutations

1 Duplication: a section occurs twice

2 Deletion: a piece of DNA is removed

3 Inversion: breaks occur in a chromosome and the broken piece joins up the wrong way around

4 Translocation: part of a chromosome breaks off and reattaches to the wrong chromosome

5 Non-disjunction: incorrect separation of the chromosome during meiosis, results in aneuploidy, incorrect number of chromosomes in daughter cells.

Mutants

**MUTATIONS**

*Answer:* A change in the DNA when the DNA molecule is copied during mitosis or meiosis, or when the chromosomes separate in meiosis.

*Answer:* The mutation occurs in the somatic / body cells of the individual. Only the individua is affected by the mutation. E.g. cancerous growths

*Answer:*

A recessive allele that, when inherited in the homozygous condition results in the death of the embryo, foetus or child.

*Answer:* Due to a change in a single nucleotide, only one base is changed. May not change the amino acid sequence, and could result in a silent mutation.

*Answer:* The mutation occurs in the reproductive cells and the gametes produced. As such the mutation is inherited. E.g. PKU

*Answer:*

Results from non-disjunction of the 21st chromosome. People with Down Syndrome have trisomy-21.

Characteristics include: a characteristic facial expression, intellectual disability and weak muscles, they may also have heart defects or digestive abnormalities.

*Answer:*

Results from a point mutation in the HBB gene that codes for one of the beta-globulin proteins that make up haemoglobin the red blood cells.

Results in one different amino acid (valine instead of glutamic acid) which results in the red blood cell having a distorted or sickle shape.

Heterozygotes for the condition – have sickle-cell trait, and have no ill effects, unless in a low oxygen environment.

Homozygotes for the condition – have sickle-cell anaemia and usually die before reproducing.

Sickle-cell allele provides a selective advantage in areas with malaria and is maintained in these populations.

**Case study: Sickle-cell anaemia**

**Case study: Down Syndrome**