MUTATION

INQUIRY QUESTION 1:

How does mutation introduce new alleles into a population?

- **Explain how mutagens operate, including but not limited to:**
  - **electromagnetic radiation sources**
  - **chemicals**
  - **naturally occurring mutagens**

MUTAGENS AND MUTATIONS

A mutation is a change in the sequence of _____ in DNA. Mutations can be as small as a change to one base in the DNA sequence, or as large as changes in whole sets of chromosomes. The size of the mutation, however, has no bearing on how large the

_____ on the organism will be. Sometimes, very small changes can have devastating effects, and quite large changes can be relatively inconsequential. Mutations sometimes do not affect the functioning of the organism at all. Some mutations are spontaneous, often due to mistakes during the duplication of DNA and some can be caused by mutagenic chemicals, radiation or naturally occurring mutagens.

_____ are agents that cause mutagenic change. Although many mutations are spontaneous, usually occurring as errors during cell division when the DNA is being copied, some can be triggered by mutagens. Mutations in cells are also the cause of cancer. Cancer happens when cells divide without control mechanisms, and proliferate to such a degree that they interfere with the normal functioning of the body. These cells are altered so that they no longer respond to the controls that the body normally has over cells. They are also cells that are immortal in the sense that they can go on dividing indefinitely.

ELECTROMAGNETIC RADIATION

Mutagens can be types of ionising radiation, such as gamma rays, X-rays, UV rays.

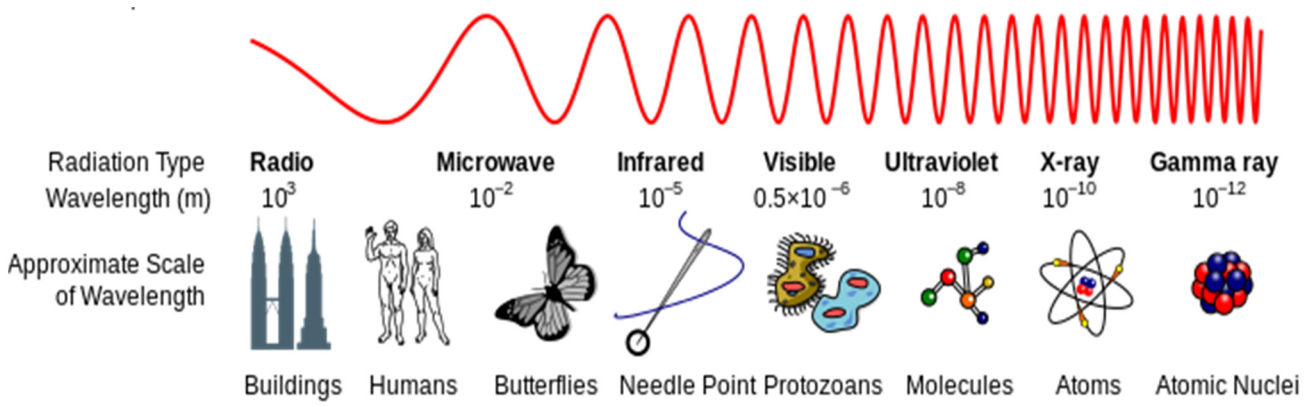

Image 1: The Electromagnetic spectrum - harmful ionising radiation has a short wavelength

Gamma rays

Gamma rays in background radiation come mostly from naturally occurring radioactive _____ of uranium, thorium and potassium in the environment. Most people are exposed to doses that are generally very small but exposure has been linked to cancers such as leukaemia. Radiation and particles given off by radiation can damage DNA. The extent of the damage upon exposure of small doses is hard to quantify. Large scale exposure to other forms of high energy radiation, such as the atomic bombs of Hiroshima and Nagasaki, and the nuclear accident at Chernobyl, have taught us that populations exposed to these types of radiation have higher incidences of cancer and birth defects than the rest of the population.

X-rays

In 1926, Hermann Muller found that if he exposed fruit flies to X-rays their mutation rates increased greatly. The mutant flies that he produced proved useful in genetic studies, and his work also showed that the changes in DNA were at the chemical level. Muller also realised that the cells of humans were vulnerable to _____ by X-rays as well. People who worked with X-rays had higher rates of _____ than the rest of the population, and this was due to mutations in their body cells that brought about cancerous changes in the tissues. Epidemiology has shown the links between X-rays and cancer.

UV waves

In 1928, Lewis Stadler produced mutations in fruit flies with ultra violet light. All UV light is considered dangerous. UVB waves damage skin and cause skin cancers and UVA are thought to enhance these effects. UVC waves are mostly trapped by the ozone layer but are used to kill bacteria. UVB waves cause disruption to bacterial cellular processes and UVC with a directly damages their DNA. It was later shown that the mutations occurred because the thymine bases of adjacent DNA strands were forming cross linkages which causes the strands to distort, which interfered with their replication.

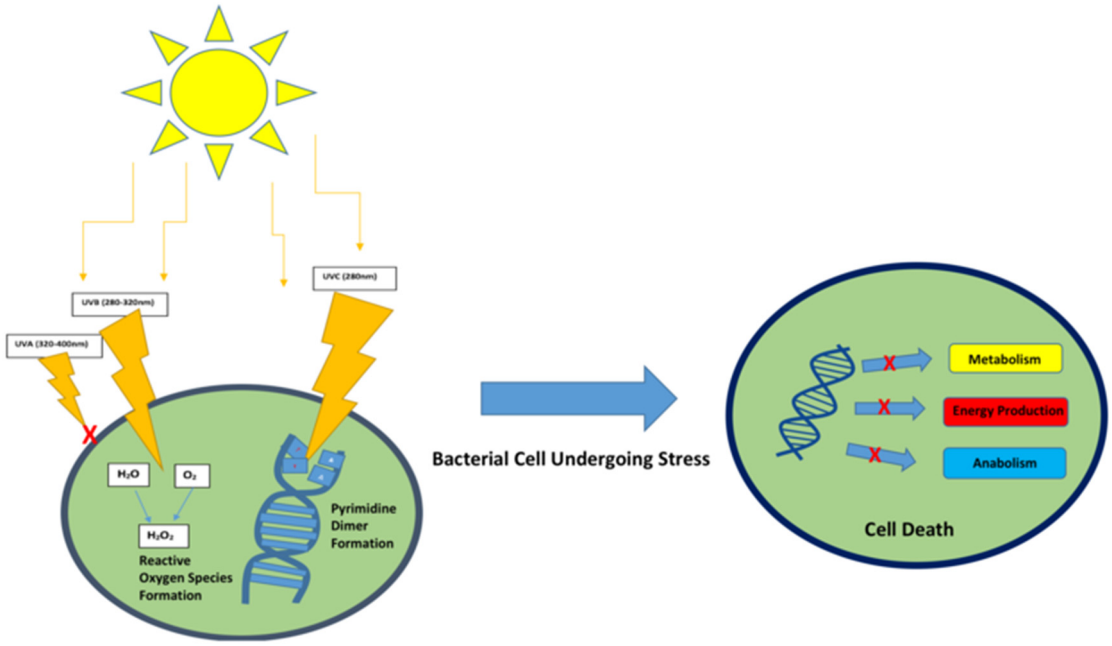

Image 2: UVB and UVC waves penetrate the bacterial cell wall and cause damage

QUESTION 1
In the 1920's dentists when X-raying teeth, used to stand above the patient and hold the X-ray film in the patient's mouth with their thumbs exposed. Today, the dentist and his assistant leave the room after placing the X-ray film in a holding frame and placing a lead shield over the patient's body. Explain why such precautions are now taken.

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CHEMICAL MUTAGENS

Certain types of chemicals in the environment can increase the rate of mutations. Mutagenic chemicals include substances such as benzene, mustard gas, ethidium bromide, nitrous acid, formaldehyde, and many of the chemicals in cigarette smoke.

These chemical mutagens can:

- cause chemical changes to particular bases.
- disrupt cell division and lead to chromosomal errors.
- mimic the shape of normal bases and hence disrupt the DNA replication process.
- insert themselves into the bonds between base pairs, known as _____, which can lead to mutations when the DNA replicates. Such events are thought to lead to cancer.
- cause direct damage to chromosomes which may lead to breaks.

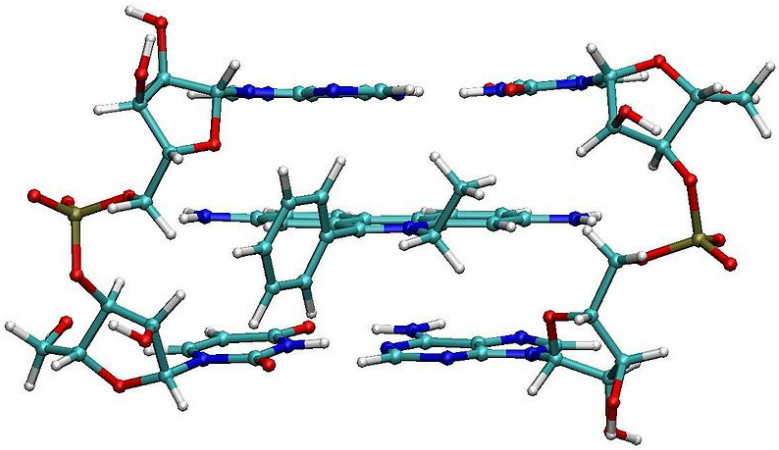

Image 3: Intercalation of the mutagen ethidium bromide (centre) between bases

Epidemiology has shown the links between smoking and lung cancer. People who don't smoke can still acquire lung cancer, but they are much less likely to do so.

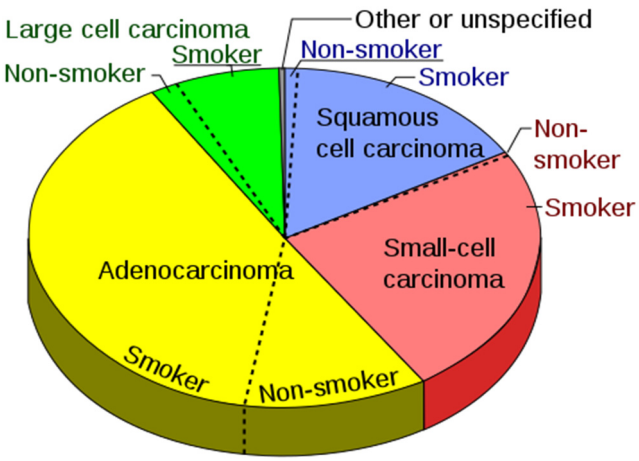

Image 4: Incidence of different lung cancers for smokers and non-smokers

NATURALLY OCCURING MUTAGENS

TRANSPOSONS

A transposable genetic element, or a transposon, is a gene or a part of a gene that is able to move from one part of a chromosome to another part of a chromosome. They are also nicknamed 'jumping genes' or mobile genetic elements. The movement of the transposon is called transposition.

Transposons make copies of themselves and then cut and paste the copies into other sites in the genome. Due to this wandering nature, transposons can insert themselves into genes and disrupt their _____. In the petunia flower below, transposons have inserted themselves into the gene for pigmentation. This gene has been interrupted and the production of pigmentation is blocked.

Image 5: Transposons have interrupted the function of the pink pigmentation gene

Transposons can also cause damage to the gene by leaving a gap when it moves. This gap may not be repaired. If transposons make multiple copies of themselves they can lengthen the gene which may lead to mis-pairing during mitosis or meiosis, this in turn affects crossing over.

QUESTION 2
Sequences of DNA that move from one position to another position in eukaryotic cells are known as:

A Jumping chromosomes
B Transposons
C Crossing over genes
D Restriction enzymes

QUESTION 3
Using an example, describe how transposable genetic elements (transposons) can disrupt a gene.

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