Mutations

Mutations are changes in genes, which are passed on to daughter cells. Normally replication is extremely accurate but very occasionally mistakes do occur. Changes in DNA can lead to changes in cell function like this:



There are three kinds of gene mutation, shown in this diagram:



Read the following text about mutations. Highlight key words.

Write five questions on this text, swap them with your partner to answer each other’s questions.

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| Effect of mutations:1. A nonsense mutation occurs if the base change results in the formation of a stop codon. This would mean that the production of the polypeptide would be stopped prematurely and the protein would be unlikely to function.
2. A silent mutation occurs when the substituted base still codes for the same amino acid as before – this is because the genetic code is degenerate.
3. A mis-sense mutation occurs when the base change results in a different amino acid being coded for. The effect this has depends on several factors:
* If a single amino acid is changed to a similar one (e.g. both small and uncharged), then the protein structure and function may be unchanged, but if an amino acid is changed to a very different one (e.g. an acidic R group to a basic R group), then the structure and function of the protein will be very different.
* If the changed amino acid is at the active site of the enzyme then it is more likely to affect enzyme function than if it is part of the supporting structure.
1. Additions and Deletions are Frame shift mutations and are far more serious than substitutions because more of the protein is altered.
* If a frame-shift mutation is near the end of a gene it will have less effect than if it is near the start of the gene.
* If the mutation is in a gene that is not expressed in this cell (e.g. the insulin gene in a red blood cell) then it won't matter.
* If the mutation is in a non-coding section of DNA then it probably won't matter.
* Some proteins are simply more important than others. For instance non-functioning receptor proteins in the tongue may lead to a lack of taste but is not life-threatening, whereas non-functioning haemoglobin is fatal.
* Some cells are more important than others. Mutations in somatic cells (i.e. non-reproductive body cells) will only affect cells that derive from that cell. Mutations in germ cells (i.e. reproductive cells) will affect every single cell of the resulting organism as well as its offspring. These mutations are one source of genetic variation.

As a result of a mutation there are three possible phenotypic effects.* Most mutations have no phenotypic effect. These are called silent mutations, and we all have a few of these.
* Of the mutations that have a phenotypic effect, most will have a negative effect. When an enzyme stops working, a metabolic block can occur, when a reaction in cell doesn't happen, so the cell's function is changed. An example of this is the genetic disease phenylketonuria (PKU), caused by a mutation in the gene for the enzyme phenylalanine hydroxylase. This causes a metabolic block in the pathway involving the amino acid phenylalanine, which builds up, causing mental retardation.
* Very rarely a mutation can have a beneficial phenotypic effect, such as making an enzyme work faster, or a structural protein stronger, or a receptor protein more sensitive. Although rare beneficial mutations are important as they drive evolution.

The kinds of mutations discussed so far are called point or gene mutations because they affect specific points within a gene. There are other kinds of mutation that can affect many genes at once or even whole chromosomes.  |

Read about chromosome mutations on page 221 and summarise the two different forms:

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Mutagens

The rate of mutations is increased by chemicals or by radiation. These are called mutagenic agents or mutagens, and include:

* High energy ionising radiation such as x-rays, ultraviolet rays, alpha, beta, or gamma rays from radioactive sources. These ionise the bases so that they don't form the correct base pairs.
* Intercalating chemicals such as mustard gas (used in WW1), which bind to DNA separating the two strands.
* Chemicals that react with the DNA bases such as benzene, nitrous acid, and tar in cigarette smoke.
* Viruses. Some viruses can change the base sequence in DNA causing genetic disease and cancer.

These mutagens lead to acquired mutations, where a cell’s DNA is damaged over time due to environmental influences. This is different from hereditary mutations, where a cell’s DNA carries a change that has been passed down from the mother or father.

Meiosis

Sex cells are …………………………………… which means they have only one of each pair of ………………………………………… …………………………………………… When a sperm (2n/n) fertilises an egg (2n/n) a ……………………………………………… is formed (2n/n).

Meiosis consists of (one/two) divisions. Before meiosis starts, each chromosome ………………………………………… to form two ……………………………………

Homologous chromosomes come together to form ……………………………………

During the first division, the …………………………………… divide. Sections of chromatids can be swapped between homologous chromosomes – this is called ………………………………… ……………………………………… and leads to ………………………………………… It happens at points called …………………………………………

During the second division, the ………………………………… separate producing ………………………… daughter cells with alternative combinations of chromosomes – this is called ………………………………………… ………………………………………………

3 ways in which meiosis leads to variation:

1. Crossing over

1. Independent segregation

In this simple example with 2 homologous chromosomes (n=2) there are 4 possible different gametes (22). In humans with n=23 there are …………………………………. possible different gametes.

1. Random fertilisation – any female gamete can join with any male gamete – so every zygote is unique.

Because meiosis leads to variation in offspring, some may be better adapted to survive than others. These successful organisms then pass on their genes to their offspring.

Similarities and differences between mitosis and meiosis

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Now make a glossary sheet for this topic.

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| **Cause of variation**  | **How variation is produced**  |
|  | An error occurs during replication of the DNA in a gene. A change in the order of base alters the amino acid sequence in the protein coded for by the gene.  |
|  | During meiosis maternal and paternal chromosomes are reshuffled. The chromosomes and therefore alleles of genes can combine in new ways.  |
|  | During metaphase 1 sections of chromatids in the bivalents are interchanged. Blocks of genes are moved and linked alleles may separate and rejoin in new combinations.  |
|  | During cell division sections of chromosomes are displaced, e.g. during anaphase. This can result in genes being deleted, duplicated or inversion of a sequence.  |
|  | Each parent is genetically different and can produce huge numbers of gametes. Which gametes fuse at fertilisation is a matter of chance.  |
|  | The expression of genes may be affected by diet, disease or temperature during development. Mutagens may cause gene mutations in somatic cells.  |

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| Further reading and questionsChapters 9.1 and 9.2. Try summary questions and look at application box starting p. 221.\*\*\*maths worked example page 228 then try qu. 5 in orange box\*\*\* |