STRUCTURAL CHROMOSOMAL ABERRATIONS

Structural chromosomal aberrations cause structural abnormalities in chromosome structure. They alter the sequence or the kind of genes present in chromosome. These are further classified into four groups based upon whether they alter the gene sequences, number or location.

Changes in the structure of chomosomes.

a. Loss or addition of segments of chromosomes.

Deletion (deficiency) - Loss of a segment of a chromosome

Duplication - repetition of a segment of a chromosome.

b. Changes in the normal arrangement of genes in the chromosome.

Translocation – Exchange of segments between two non - homologous chromosomes, resulting in new chromosomes.

Inversion – Change in the linear order of genes by rotation of a section of a chromosome through 180 degrees.

Gene mutations or point mutations – changes in the nucleotide sequence of a gene.

a. Deletion
b. Insertion
c. Substitution
d. Inversion

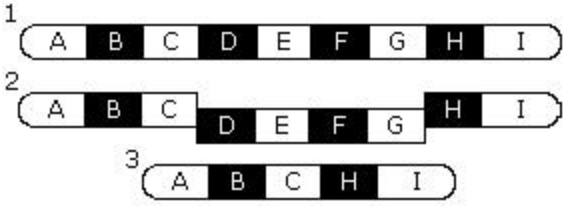
a. Loss or addition of segments of chromosomes

Deletion of a Gene

As the name implies, genes of a chromosome are permanently lost as they become unattached to the centromere and are lost forever

Normal chromosome before mutation

- Genes not attached to centromere become loose and lost forever
- New chromosome lacks certain genes which may prove fatal depending on how important these genes are



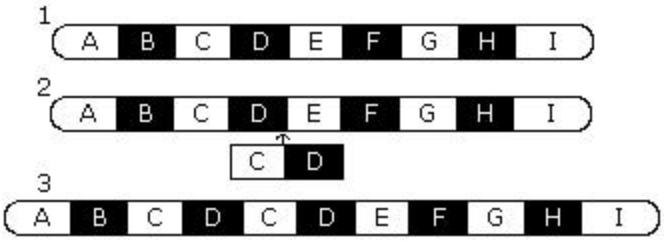
Duplication of Genes

In this mutation, the mutants genes are displayed twice on the same chromosome due to duplication of these genes. This can prove to be an advantageous mutation as no genetic information is lost or altered and new genes are gained

Normal chromosome before mutation

- Genes from the homologous chromosome are copied and inserted into the genetic sequence
- New chromosome possesses all its initial genes plus a duplicated one, which is usually harmless

The next page continues looking at these chromosome mutations and mutations that happen within genes that can prove to be more harmful to the organism at hand. The following pages also investigates polyploidy in species.

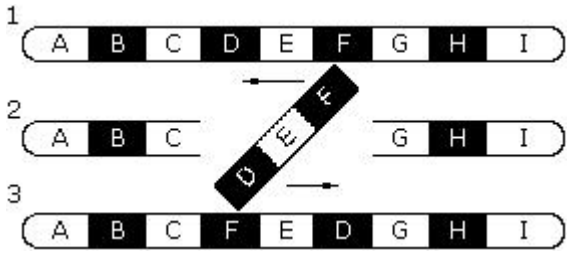


b. Changes in the normal arrangement of genes in the chromosome

Inversion of Genes

This is where the order of a particular order of genes are reversed as seen below

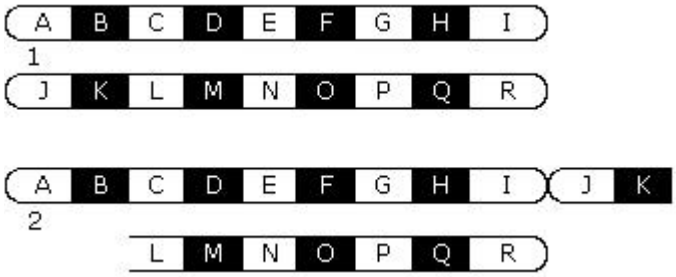
- Normal chromosome un-altered
- The connection between genes break and the sequence of these genes are reversed
- The new sequence may not be viable to produce an organism, depending on which genes are reversed. Advantageous characteristics from this mutation are also possible



Translocation of Genes

This is where information from one of two homologous chromosomes breaks and binds to the other. Usually this sort of mutation is lethal

- An un-altered pair of homologous chromosomes
- Translocation of genes has resulted in some genes from one of the chromosomes attaching to the opposing chromosome



II. Gene mutations or point mutations

Alteration of a DNA Sequence

The previous examples of mutation have investigated changes at the chromosome level. The sequence of nucleotides on a DNA sequence are also susceptible to mutation.

Substitution

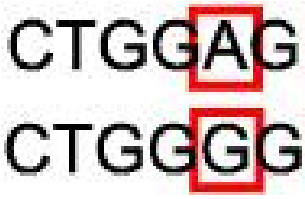
A substitution is a mutation that exchanges one base for another (i.e., a change in a single "chemical letter" such as switching an A to a G). Such a substitution could:

1. change a codon to one that encodes a different amino acid and cause a small change in the protein produced. For example, sickle cell anemia is caused by

a substitution in the beta-hemoglobin gene, which alters a single amino acid in the protein produced.

2. change a codon to one that encodes the same amino acid and causes no change in the protein produced. These are called silent mutations.

change an amino-acid-coding codon to a single "stop" codon and cause an incomplete protein. This can have serious effects since the incomplete protein probably won't function.



Insertion

Insertions are mutations in which extra base pairs are inserted into a new place in the DNA.

Deletion

Deletions are mutations in which a section of DNA is lost, or deleted.

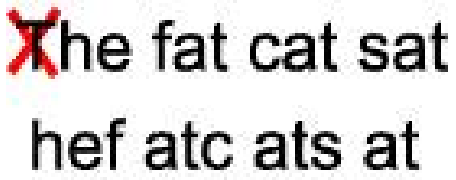
Inversion

Frameshift

Since protein-coding DNA is divided into codons three bases long, insertions and deletions can alter a gene so that its message is no longer correctly parsed. These changes are called frameshifts.

For example, consider the sentence, "The fat cat sat." Each word represents a codon. If we delete the first letter and parse the sentence in the same way, it doesn't make sense.

In frameshifts, a similar error occurs at the DNA level, causing the codons to be parsed incorrectly. This usually generates truncated proteins that are as useless as "hef atc ats at" is uninformative.



Genetic mutations increase genetic diversity and therefore have an important part to play. They are also the reason many people inherit diseases.