Mutation Notes

Mutations- changes to genetic material.

Two types: **Gene Mutations** and **Chromosomal Mutations**

Gene Mutations (a.k.a. point mutations)- changes to a single gene

Point mutations- changes in one or a few nucleotides at a single point in the DNA sequence. ex. substitutions, insertions, deletions.

Insertions and deletions are ***Frameshift Mutations*** and tend to be more problematic than substitutions. Substitutions tend to result in one wrong amino acid in the chain. Frameshift Mutations alter the codons read from that point on, therefore the entire sequence of amino acids is incorrect. This results in an altered protein that is unable to perform its normal function.

Chromosomal Mutations

Mutations involve in the changes of the number or structure of chromosomes. These changes can affect the structure, function, and inheritance of whole DNA molecules (microscopically visible in a coiled state as chromosomes). Often these chromosome mutations result from one or more coincident breaks in the DNA molecules of the genome (possibly from exposure to energetic radiation), followed in some cases by faulty rejoining. Some outcomes are large-scale deletions, duplications, inversions, and translocations. In a diploid species (a species, such as human beings, that has a double set of chromosomes in the nucleus of each cell), deletions and duplications alter gene balance and often result in abnormality. Inversions and translocations involve no loss or gain and are functionally normal unless a break occurs within a gene. However, at meiosis (the specialized nuclear divisions that take place during the production of gametes—i.e., eggs and sperm), faulty pairing of an inverted or translocated chromosome set with a normal set can result in gametes and hence progeny with duplications and deletions.

Loss or gain of whole chromosomes results in a condition called aneuploidy. One familiar result of aneuploidy is Down syndrome, a chromosomal disorder in which humans are born with an extra chromosome 21 (and hence bear three copies of that chromosome instead of the usual two). Another type of chromosome mutation is the gain or loss of whole chromosome sets. Gain of sets results in polyploidy—that is, the presence of three, four, or more chromosome sets instead of the usual two. Polyploidy has been a significant force in the evolution of new species of plants and animals.

many mutations are silent, showing no obvious effect at the functional level. Some silent mutations are in the DNA between genes, or they are of a type that results in no significant amino acid changes.

Mutations: good or bad?

Many, if not most, mutations are neutral to gene expression. Mutations that cause dramatic changes in protein structure or gene activity often disrupt normal biological activities. ex. sickle-cell anemia, cystic fibrosis. Beneficial mutations produce genetic variation which can be beneficial to populations. New proteins or altered activity can be useful in a changing environment. ex. HIV resistance.

Different types of effects of mutation

Effects of Deletion

- Small deletions are not fatal but homozygous deletions are lethal. They cause the abnormal development.
- Framshift mutation causes the production of non-functional protein.
- Male infertility.
- Cry of the cat syndrome.

Effects of Duplication

- Role in evolutionary biology

Effects of Inversion

- Production of unbalance chromosome rearrangements in offspring.
- Lead to an increased risk of miscarriage due to effects on chromosome 9.

Effects of Translocation

- Cancer
- Infertility
- Down's syndrome