Mutations Notes

What is a mutation?
Changes in the genetic material (DNA).

Mutations can occur in two different types of cells:
1. Somatic (body) cells
2. Gamete (sex) cells

Mutations that occur in somatic (body) cells usually result in killing that body cell only.
An exception to this is if the mutation occurs to the DNA that controls regulation of the cell cycle. This can result in cancer.

- Somatic mutations usually only affect the individual.
- Somatic mutations can NOT be passed on to the next generation.

Mutations that occur in gamete (sex) cells means that every cell of the developing fetus/baby will have that mutation.

- Gamete mutations usually do affect the individual with some genetic disorder.
- Gamete mutations can be passed on to the next generation.

There are two types of mutations that can occur in gamete cells:
1. Gene Mutations
2. Chromosomal Mutations

2 types of Single Gene Mutations:

1. Point Mutation: a single point is changed in the DNA sequence.

THE FAT CAT ATE THE **R**AT

THE FAT CAT ATE THE **H**AT

- Can be a substitution in which one base is changed into another base.

DNA: TAC GCA TGG AAT
mRNA: AUG C**G**U ACC UUA
Amino Acid: Met - **Arg** - Thr - Leu

DNA: TAC G**T**A TGG AAT
mRNA: AUG C**A**U ACC UUA
Amino Acid: Met - **His** - Thr - Leu

Point mutations usually affect no more than a single amino acid.
The protein may be slightly affected or not affected at all.

2. Frameshift Mutation: a single gene or nitrogen base is deleted or added from the mRNA sequence causing a shift in the "reading frame" of the genetic message.

THE FAT CAT ATE THE RAT

T**H**E FAT CAT ATE THE RAT

T**EF ATC ATA TET HER AT**

- Can be an insertion in which one base is inserted in the DNA sequence.
- Can be a deletion in which one base is deleted in the DNA sequence.

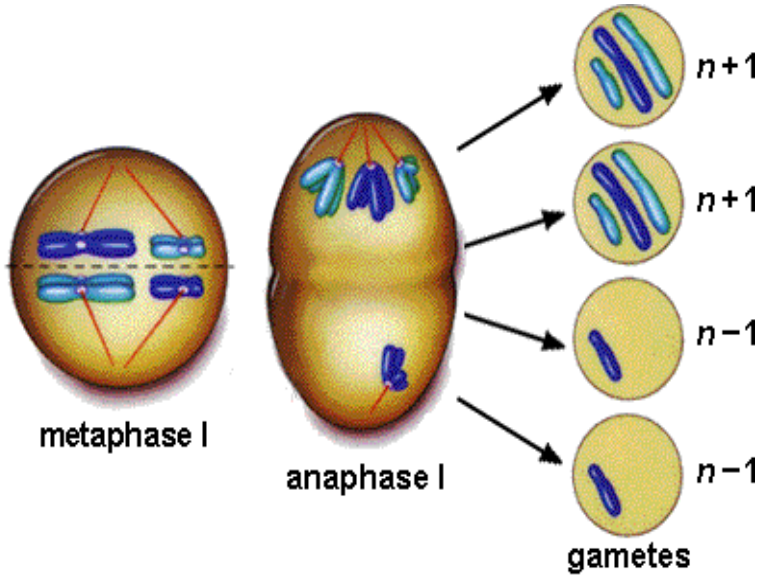
DNA: TAC GCA TGG AAT
mRNA: AUG CGU ACC UUA
Amino Acid: Met - Arg - Thr - Leu

DNA: TA**T** CGC ATG GAA T
mRNA: AU**A** GCG UAC CUU A
Amino Acid: **Iso** - **Ala** - **Tyr** - **Leu**

The affect of frameshift mutations is usually more dramatic.
- may change every amino acid that follows the point of the mutation.
- can alter a protein so much that it is unable to perform its normal functions.

5 types of Chromosomal Mutations:
1. Deletion
2. Duplication
3. Inversion
4. Translocation
5. Non-disjunction

Deletion: Involves the loss of all or part of a chromosome.
Duplication: Involves the production of extra copies of parts of the chromosome.
Inversion: Reverses the direction of parts of a chromosome.
Translocation: When one part of a chromosome breaks off and attaches to another chromosome.
Non-disjunction: Means "not coming apart". When homologous chromosomes fail to separate properly during meiosis.



- Results in abnormal numbers of chromosomes.
- Trisomy means a person has an extra copy of a chromosome.
- Monosomy means a person is missing a copy of a chromosome