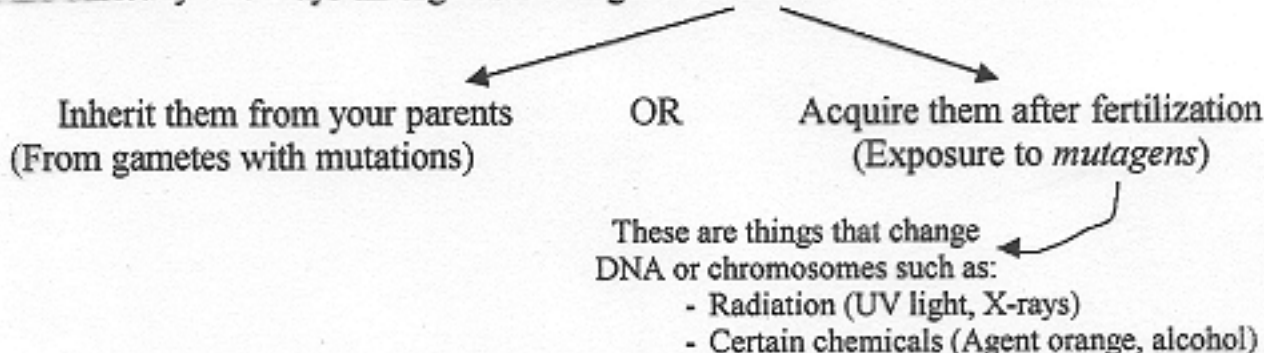


NOTHING'S PERFECT *MUTATIONS*

During replication, transcription, translation or gamete formation (AKA meiosis), enzymes are processing unthinkable numbers of base pairs. As you saw in the Genetic Police Sketch Artist lab, there are plenty of opportunities for error.

MUTATION: Any change in the genetic sequence of bases.

There are basically two ways an organism can get a mutation:



Mutations are often divided into two categories:

CHROMOSOMAL MUTATIONS: Changes that involve sections of chromosomes or even whole chromosome. These involve entire genes and often take place during meiosis.

1. **Structural changes:** sections of chromosome often break off and get turned around (*inversion*) or get added onto other chromosomes (*addition* and *translocation*) or just float away never to be transcribed again (*deletion*).
2. **Nondisjunction and Polyploidy:** During gamete formation or *meiosis*, sometimes the chromosomes do not divide evenly into the newly-divided gametes. Some may have extras while others will be short chromosomes. If these gametes go on to form a zygote, the resulting organism will have the wrong number of chromosomes. *Downs syndrome* is the result of an extra 21st chromosome.

GENE MUTATIONS: Alterations in the sequence of bases of DNA (or mRNA). These will often cause changes in amino acid sequence of the resulting polypeptide.

1. **Substitution:** When one base is replaced with another. The "safest" because they only affect one codon.
2. **Frameshift mutations:** These involve adding or deleting a base. This causes all of the codons afterward to be affected

Some Changes Go Unseen

SILENT MUTATIONS: Quite a bit of DNA does not code for anything (we think). It is often referred to as “trash” or “garbage” DNA. Most often, mutations to these sections are not expressed and cause no change in the organism.

LETHAL MUTATIONS: These alterations so affect the biochemistry of the organism that it cannot survive. About half of every conception (fertilization of an egg) involves gametes that carry lethal mutations or mutations in the early stages of development that prevent the zygote from surviving. In humans, these cells are either reabsorbed or shed with the uterine lining during menstruation.

Detecting Genetic Disorders

Many genetic disorders cannot be treated. If there is a suspicious pregnancy, oftentimes doctors will want to karyotype the baby to see if there are any abnormalities.

KARYOTYPING: Pictures are taken of the chromosomes of a cell from the embryo. These pictures are then counted, arranged in homologous pairs and their structures compared for any abnormalities. But where do we get these cells?

AMNIOCENTESIS: In mammals, the embryo develops within the uterus in a swimming pool of sorts called the amnion. The embryo is shedding cells into the amniotic fluid around it just as you are sloughing off skin cells into the air around you. During this procedure a small amount of amniotic fluid is withdrawn and the cells in it analyzed and karyotyped.

ULTRASOUND: Using the same concept that the police use to catch speeders, a “picture” of a developing fetus (or any other body tissue) can be made by measuring how sound waves directed into the body are reflected as they pass through.

A Final Note

EVOLUTION: Not all mutations are necessarily bad. Some changes may actually be improvements. If the improvement helps the organism survive longer and make more young (who might have or carry the “new and improved” gene) chances are, that trait will become more common over time and we have.....

.....our next unit: Evolution