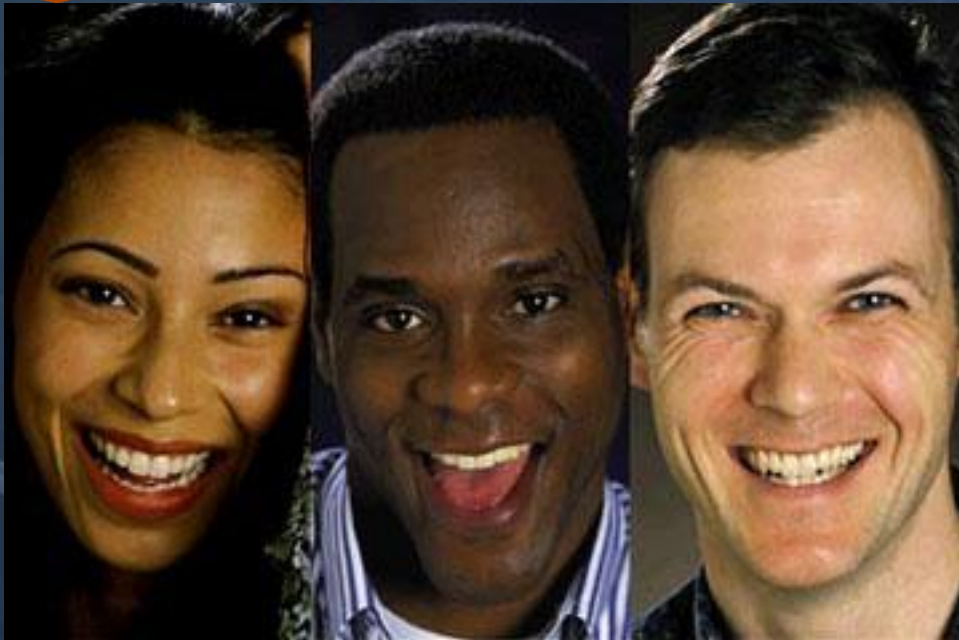


MUTATIONS



We will focus on 2 types of mutations:

- Chromosomal Mutations
- Gene Mutations



Chromosomal Mutations

- When all or parts of a chromosome change for an unknown reason.
- Usually occur during mitosis or meiosis.
- Can cause major problems because it affects many genes.
- Can be beneficial in other organisms.

Original Chromosome



Duplication



Deletion



Inversion - Paracentric



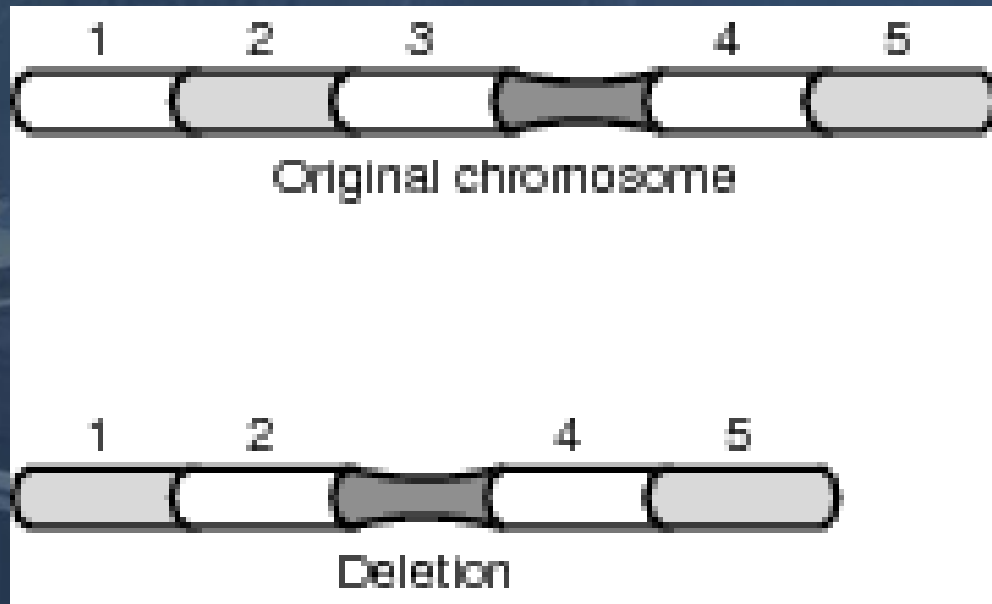
Inversion - Pericentric



Changes in Chromosomal Structure

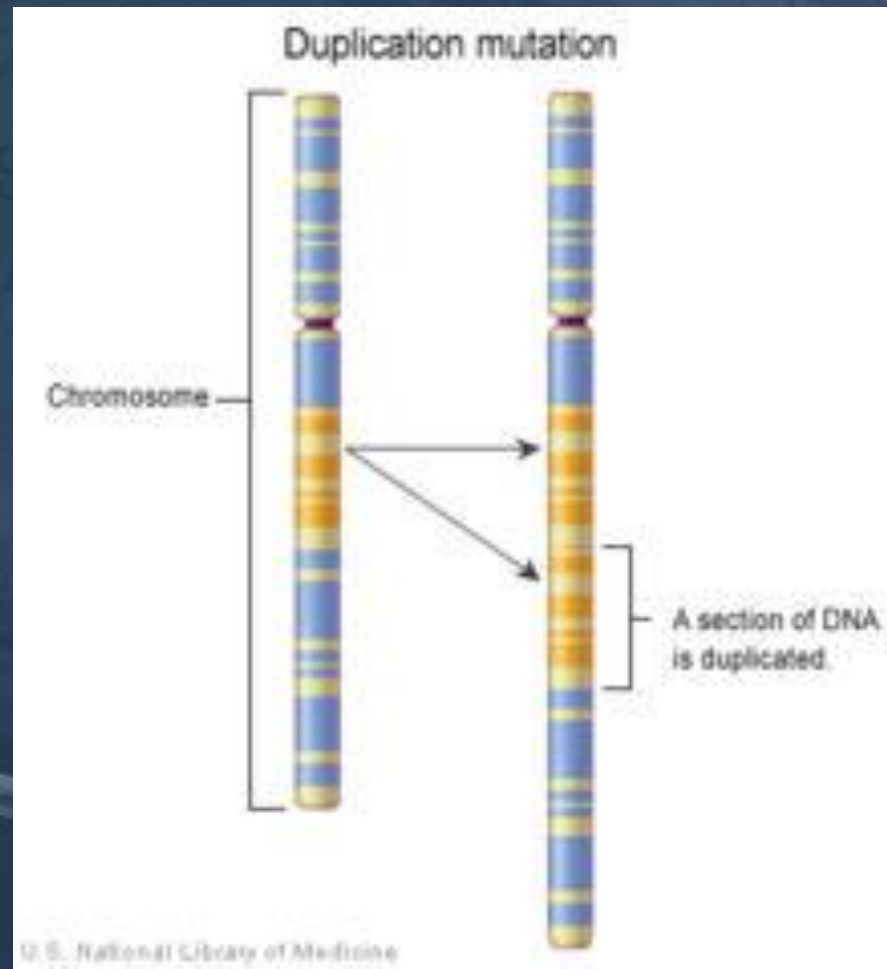
Deletion

- During the cross over in meiosis, whole parts of the chromosome may detach.
- This loses the genes on that chromosome.



DUPLICATION

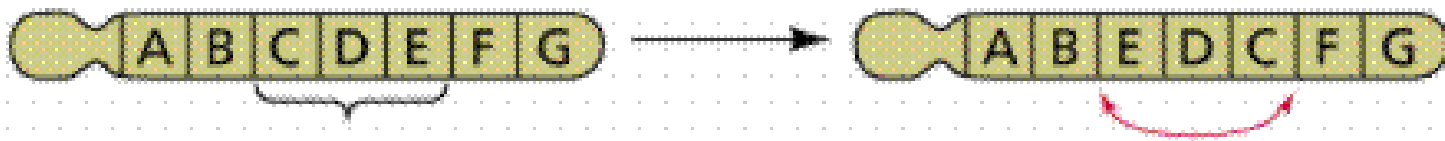
- Can also occur in meiosis during crossing over.
- The chromosome “picks up” an extra piece from homologous chromosome. Therefore genes have been duplicated.



Inversion:

- Part of chromosome breaks free then reattaches itself backwards.
- Therefore the genetic code has been rearranged.

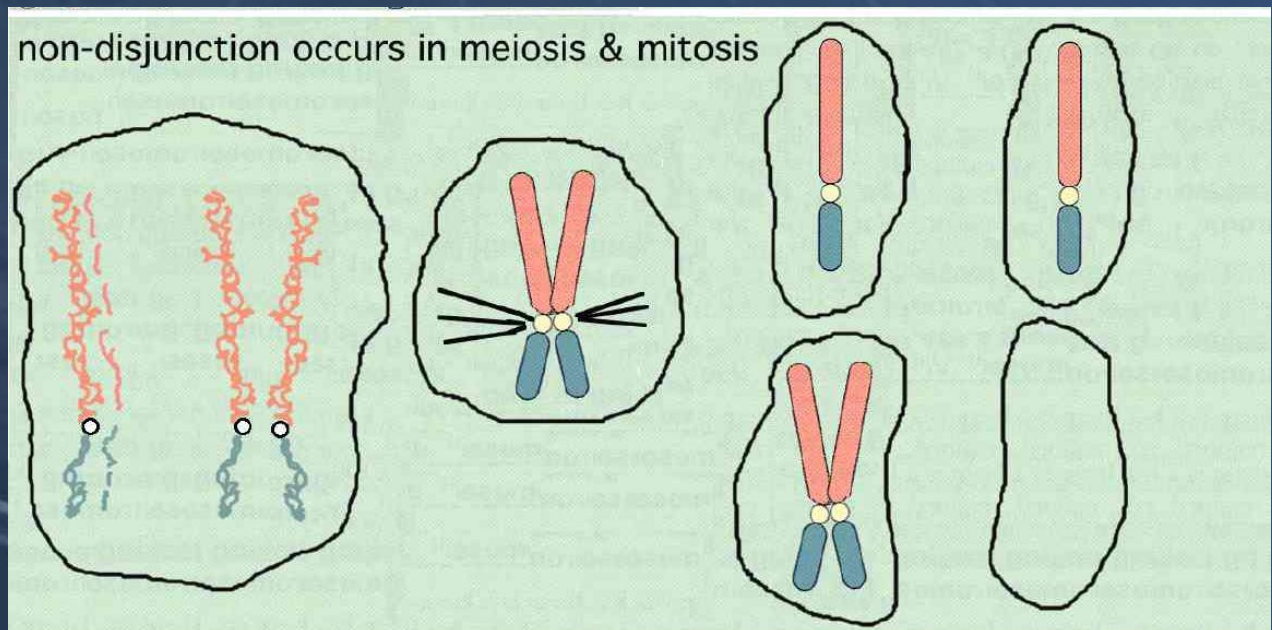
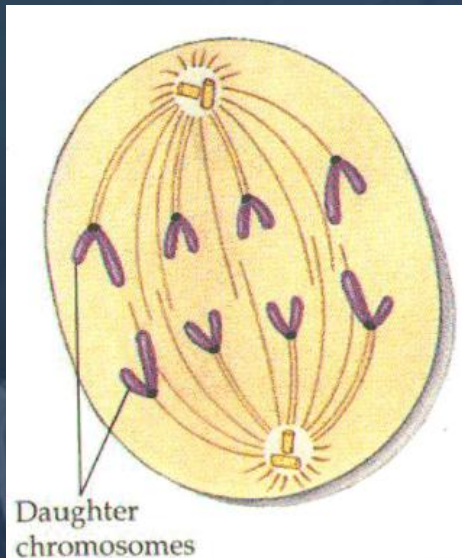
Inversion



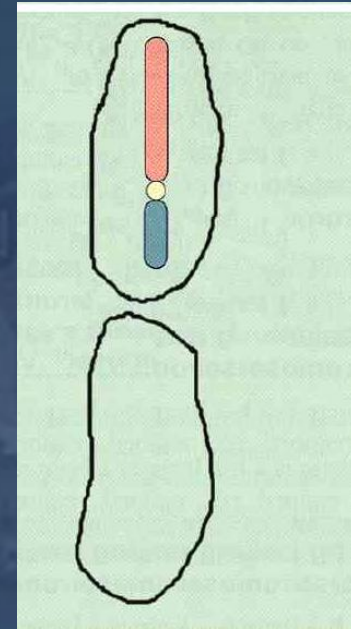
Changes in Chromosomal Number

Nondisjunction

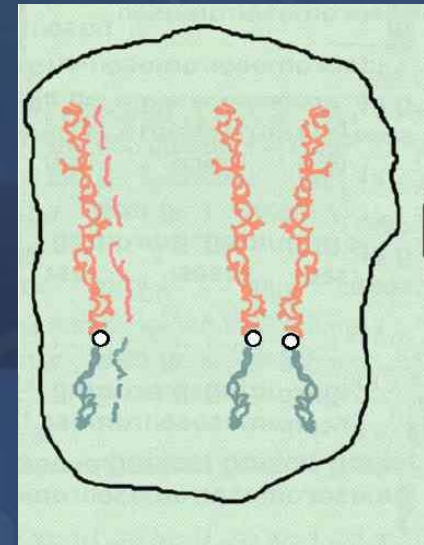
- Chromosomes fail to separate correctly during cell division
- Cell usually dies if this occurs after mitosis.
- Can be a problem if this occurs during meiosis.



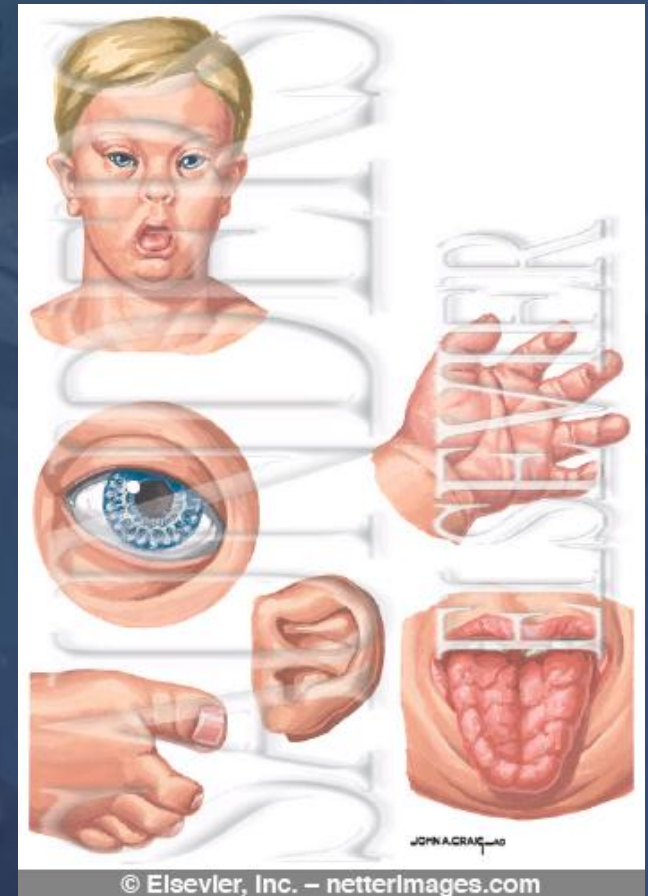
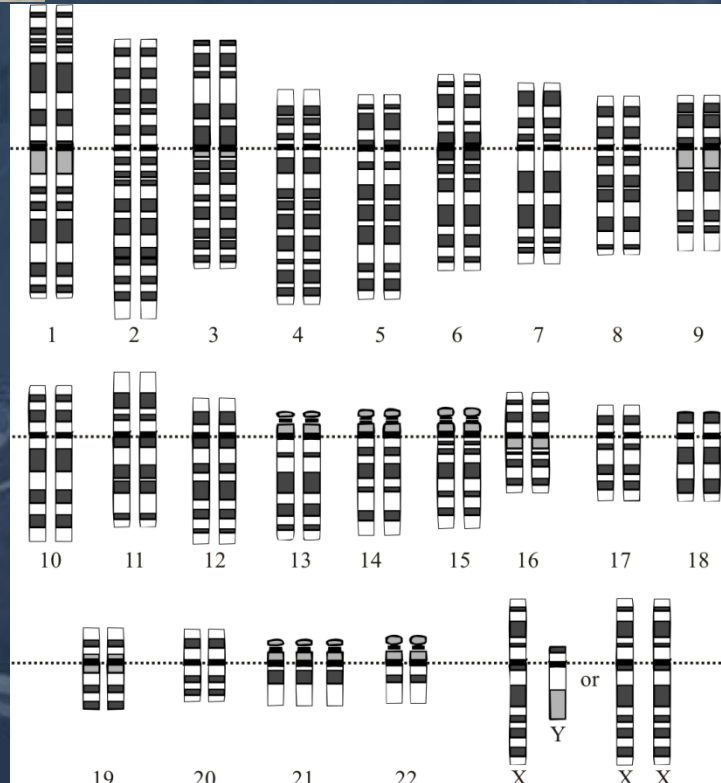
Monosomy – when the zygote forms it will be short a homologous chromosome



Trisomy – the zygote receives three homologous chromosomes



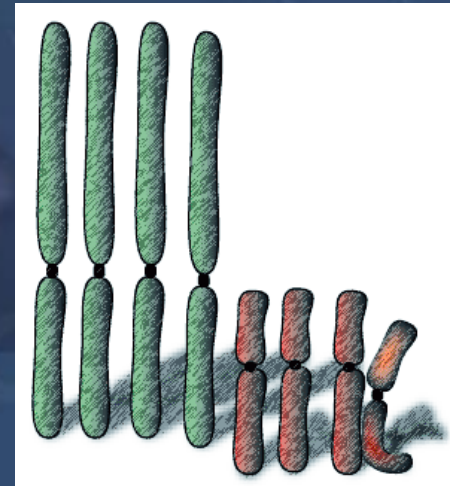
DOWN SYNDROME



*not possible in animals because they do not have the same chromosomes as humans.

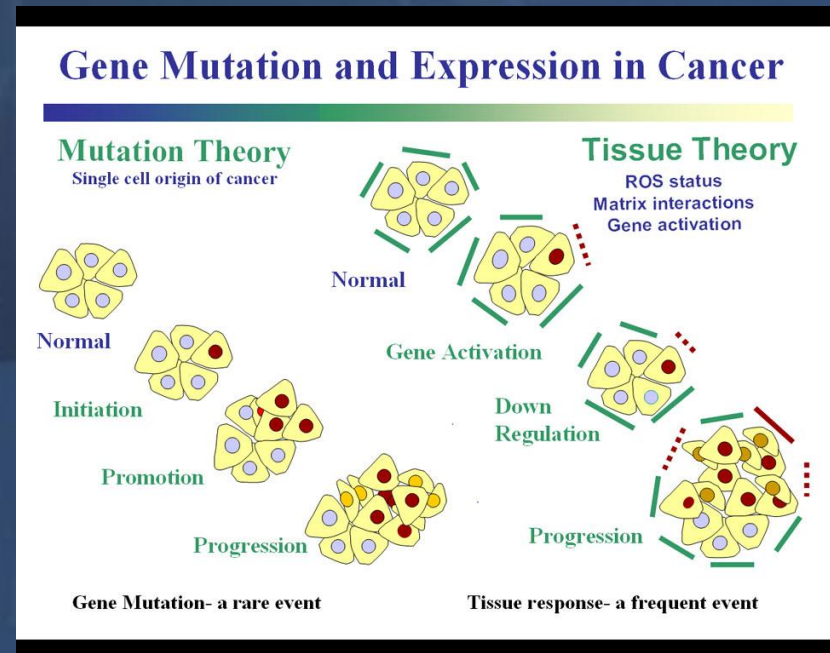
Polyploidy

- Nondisjunction occurs in all cells
- Lethal in animals, however quite common to plants.
- Can be beneficial to plants – larger flowers or fruit etc.



Gene Mutations

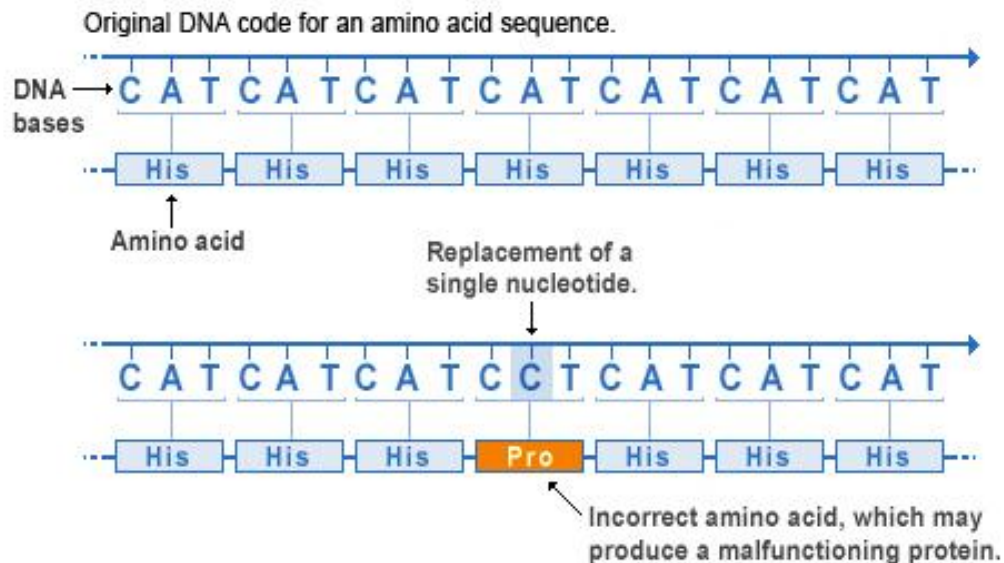
- This is a change in the nucleotide sequence
- It can be caused by:
 - DNA replication error
 - Mutagens such as UV light, radiation, chemicals, tobacco & other carcinogens



Point Mutation - Base-pair Substitution (missense, nonsense and silent)

- When a pair of bases are replaced by another base.
- Can be harmless (silent) – may code for the same amino acid
- Sickle cell hemoglobin is a harmful mutation.

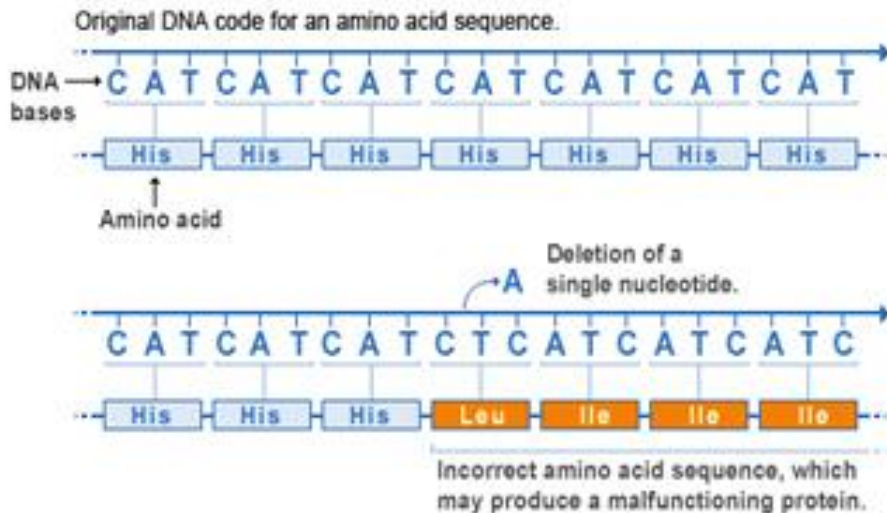
Missense mutation



Frameshift - Deletion or Addition

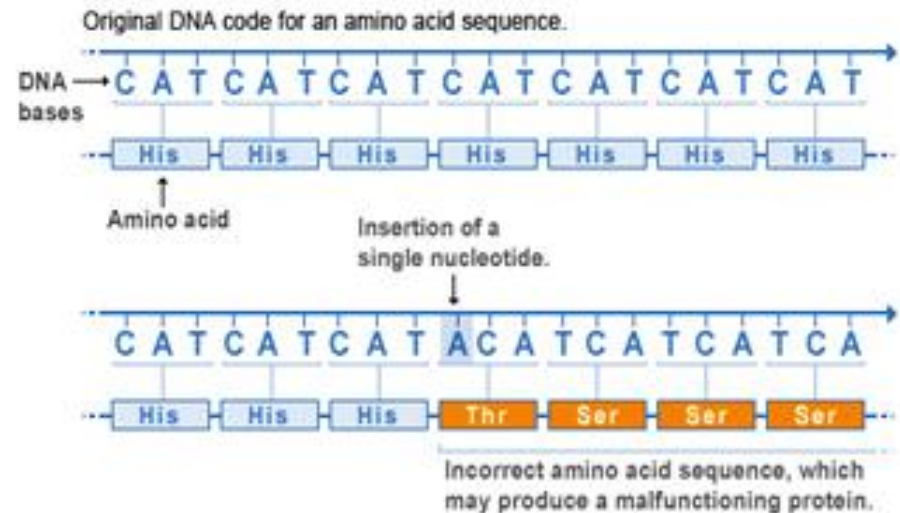
- A loss of one base.
- This can change the whole amino acid sequence

Deletion mutation



U.S. National Library of Medicine

Insertion mutation

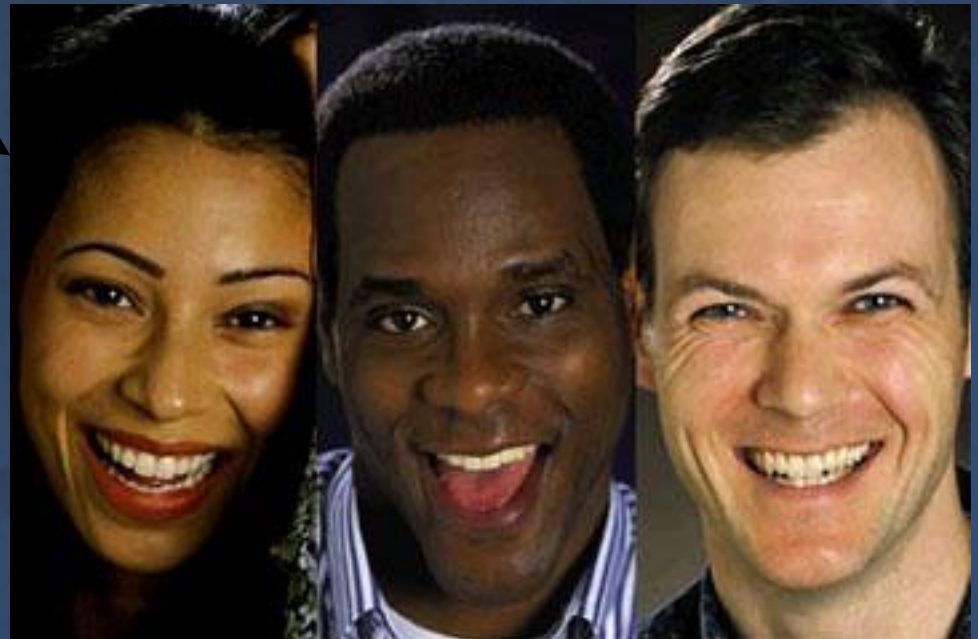


U.S. National Library of Medicine

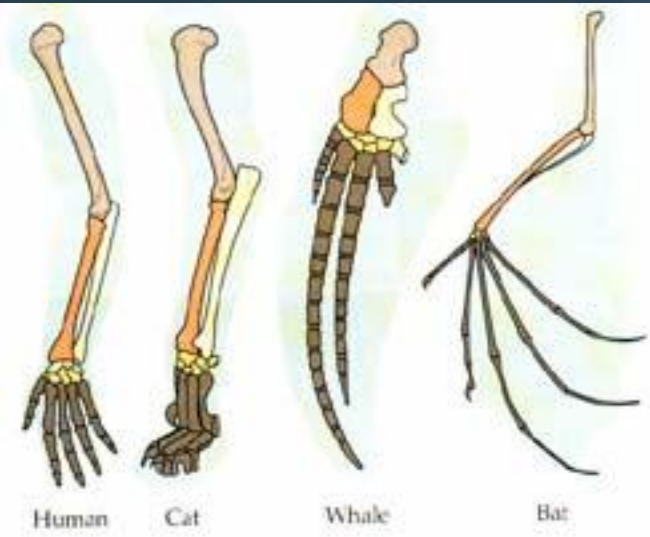
Effect of Mutations

- Death
- Disease – sickle blood cells, down syndrome
- Genetic Variation

Affect of intense UV light on organisms may affect their phenotype depending on their environment.



Natural Selection



1. Variation
2. Inheritance
3. More individuals are born that can survive
4. Variable “fitness” leads to best suited organisms surviving to reproduce

Where does the variation come from?????