

# WHAT ARE MUTATIONS?

- Changes in the nucleotide sequence of DNA
- •May occur in somatic cells (aren't passed to offspring)
- May occur in gametes (eggs & sperm) and be passed to offspring



### ARE MUTATIONS HELPFUL OR HARMFUL?

- Mutations happen regularly
- Almost all mutations are neutral
- Chemicals & UV radiation cause mutations
- •Many mutations are repaired by enzymes



### ARE MUTATIONS HELPFUL OR HARMFUL?

- •Some type of skin cancers and leukemia result from somatic mutations
- Some mutations may improve an organism's survival (beneficial)



# TYPES OF MUTATIONS



# CHROMOSOME MUTATIONS

May Involve:
Changing the structure of a chromosome
The loss or gain of part of a chromosome





## **CHROMOSOME MUTATIONS**

Five types exist:
Deletion
Inversion
Translocation
Nondisjunction
Duplication



## DELETION

# Due to breakage A piece of a chromosome is lost





### INVERSION

Chromosome segment breaks off
Segment flips around backwards
Segment reattaches





# DUPLICATION

# Occurs when a gene sequence is repeated

Duplication mutation





# TRANSLOCATION

Involves two chromosomes that aren't homologous
Part of one chromosome is transferred to another chromosomes





# NONDISJUNCTION

# Failure of chromosomes to separate during meiosis Causes gamete to have too many or too few chromosomes





#### Original Chromosome



## Duplication



### Deletion



### Inversion



### Inversion



# **GENE WUTATIONS**

- -Change in the **nucleotide sequence** of a gene
- -May only involve a single nucleotide
- -May be due to copying errors, chemicals, viruses, etc.



# TYPES OF GENE MUTATIONS

Include:
Point Mutations
Substitutions
Insertions
Deletions
Frameshift



# POINT MUTATION

- Change of a single nucleotide
- Includes the deletion, insertion, or substitution of ONE nucleotide in a gene
- Sickle Cell disease is the result of one nucleotide substitution
- Occurs in the hemoglobin gene





# FRAMESHIFT MUTATION

- **Inserting or deleting** one or more nucleotides
- Changes the "reading frame" like changing a sentence
  Proteins built incorrectly
- •Original:
  - •The fat cat ate the wee rat.
- Frame Shift ("a" added):
  - The fat caa tet hew eer at.



### AMINO ACID SEQUENCE CHANGED



