

VOCABULARY:

- **Mutation** = a random error or change in the DNA sequence that may affect whole chromosomes or just one gene.

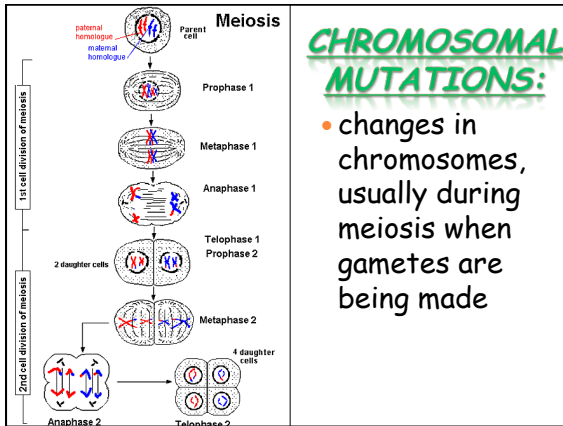
Mutations of Chromosomes

VOCABULARY:

- **Mutagen** = certain substances or conditions that can create a greater rate of mutation

MUTAGENS:

- Examples:
 - Some viruses
 - High temperatures
 - Chemicals
 - Radiation



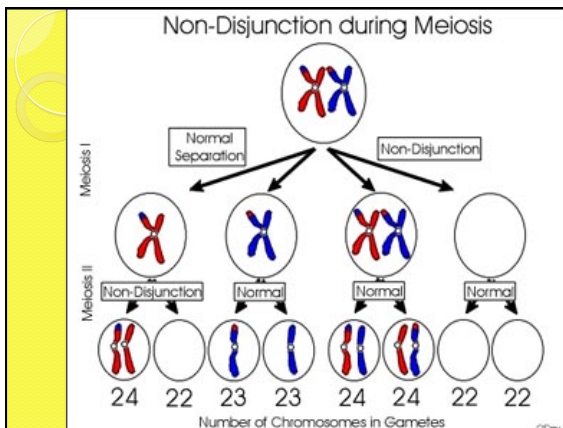
CHROMOSOMAL MUTATIONS:

- changes in chromosomes, usually during meiosis when gametes are being made

CHROMOSOMAL MUTATIONS:

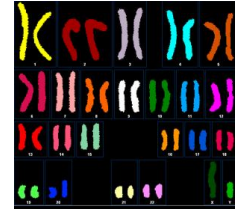
1. **Nondisjunction** = failure of homologous chromosomes to separate during meiosis resulting in gametes (egg or sperm) with too few or too many chromosomes.

- [Nondisjunction Animation](#)



CHROMOSOMAL MUTATIONS:

- **REMEMBER:** Humans are *diploid* creatures; meaning for every chromosome in our body, there is another one to match it.



CHROMOSOMAL MUTATION:

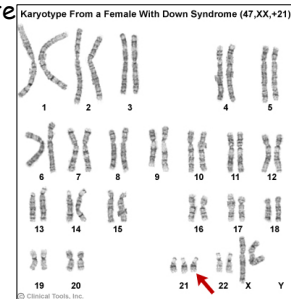
- **Aneuploidy** = abnormal number of chromosomes.

- Ex: trisomy, monosomy



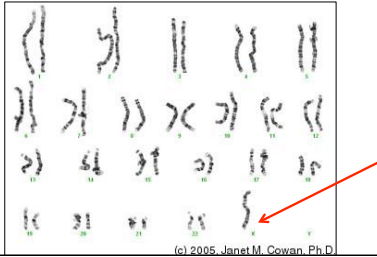
ANEUPLOIDY:

- **Trisomy** = zygote contains three copies of the chromosome.
- Ex: Down syndrome, Klinefelter's (XXY)



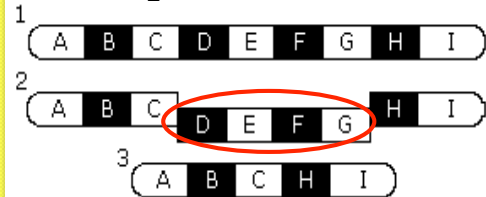
ANEUPLOIDY:

- **Monosomy** = zygote contains only one chromosome of the pair i.e. it is missing one chromosome



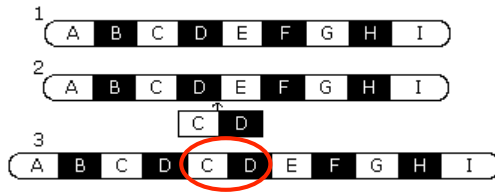
CHROMOSOMAL MUTATION:

2. **Deletion** = occurs when part of a chromosome is missing.



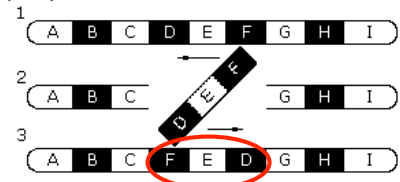
CHROMOSOMAL MUTATION:

3. **Insertion** = occurs when a part of a chromatid breaks off and attaches to its sister chromatid. The result is a duplication of genes on the same chromosome.



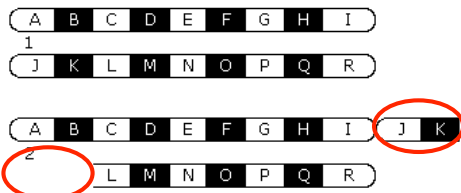
CHROMOSOMAL MUTATION:

4. **Inversion** = Segment of chromosome breaks off and is reinserted backwards (will flip upside down)



CHROMOSOMAL MUTATION:

5. **Translocation** = occurs when part of one chromosome breaks off and is added to a *different* chromosome.



GENE MUTATIONS:

- Changes in the DNA sequence that will then change the amino acid sequence. (Remember: Amino acids make up our proteins!)



GENE MUTATIONS:

1. **Point mutation** = a change in a single base pair in DNA.

GENE MUTATIONS:

Frameshift Mutation

ATG	GAA	GCA	CGT
Met	Glu	Ala	Gly

←

ATG	AAG	CAC	GT
Met	Lys	His	

2. **Frameshift mutation** = error in the DNA sequence that adds or deletes a single nitrogen base, causing nearly all amino acids following the mutation to be changed.

FRAMESHIFT MUTATION:

- Types:
 - **Base deletion** = One nitrogen base (A, T, C or G) is deleted from the DNA sequence

FRAMESHIFT MUTATION:

- **Base insertion** = Extra nitrogen base is added to the DNA sequence

FRAMESHIFT MUTATION

Frameshift mutation

Original DNA code for an amino acid sequence.

DNA bases → C A T T C A C A C G T A C T C A T G C T A T

His Ser His Val Leu Met Leu

↑
Amino acid

→ C A T T C A C A C G T A C T C A T G C T A T

Ile His Thr Tyr Ser Cys Tyr

←

Frameshift of one DNA base results in abnormal amino acid sequence.

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