



Chromosomes/DNA

Mutations

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Chromosome Mutation

- Mutations are permanent gene or chromosome changes that will be passed on to offspring if they occur in a gamete
- Two categories of chromosome mutation-Those that affect the # of chromosomes and those that affect the structure of the chromosome



ANEUPLOIDY

- Aneuploidy- an excess or deficiency of a particular chromosome.



ANEUPLOIDY

- Trisomy- occurs when an individual has an extra copy of a chromosome
- Monosomy- occurs when an individual is missing one chromosome.

TRISOMY



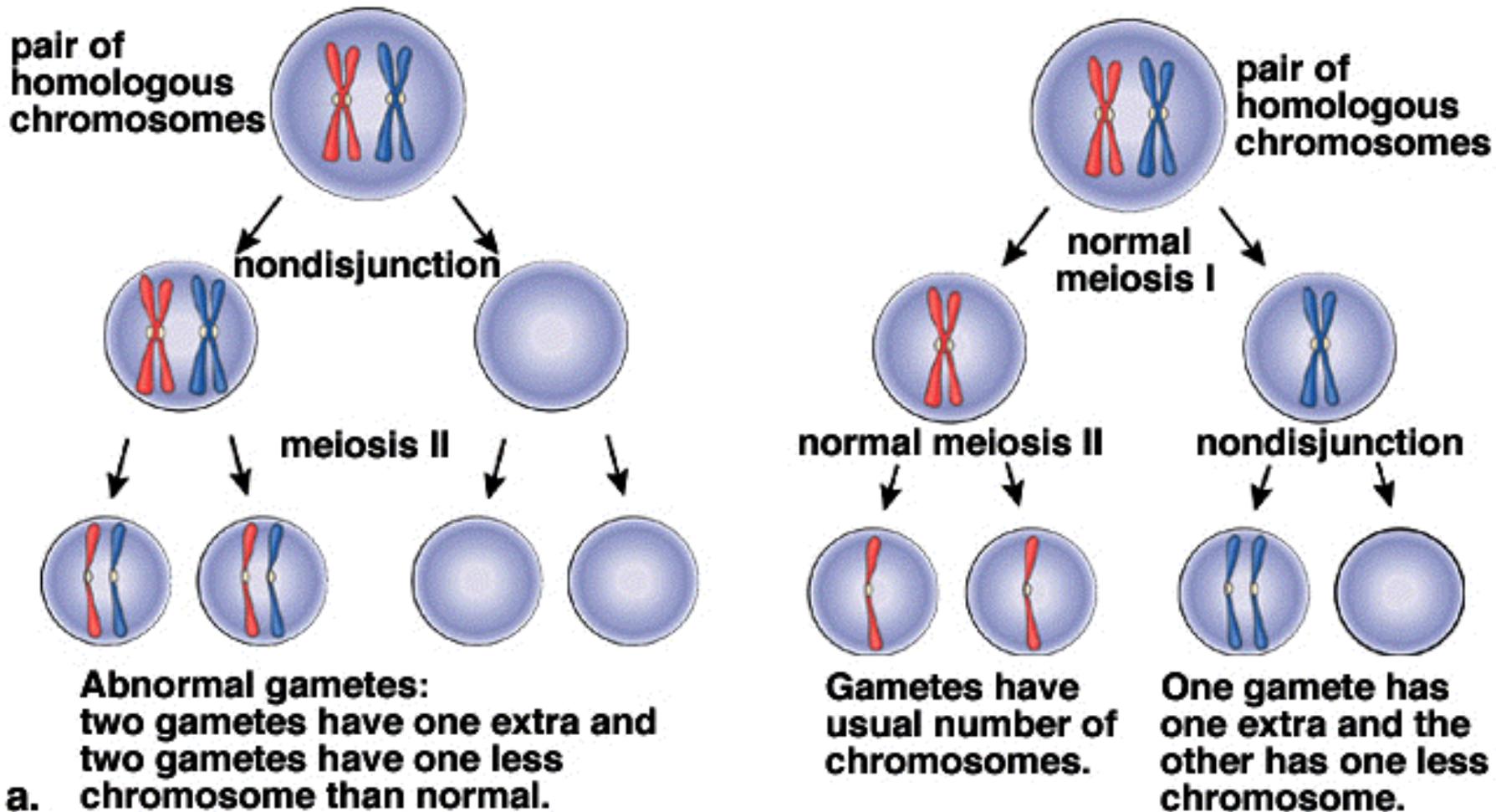
- The most common trisomy is Down Syndrome.... which is a trisomy of the 21st Chromosome.



ANEUPLOIDY

- The cause of aneuploidy is Non-disjunction- this occurs during Meiosis. When either the homologous pair (Meiosis I) or the sister Chromatids (Meiosis II) fail to separate and instead go into the same gamete.

Nondisjunction of chromosomes during meiosis





DNA Mutations

■ A Gene is a sequence of DNA nucleotide bases

■ A Gene Mutation is any change in this DNA sequence of nucleotide bases.



POINT MUTATION

- **POINT MUTATION**- a change in a single base pair.
- Sometimes this can cause no change. Sometimes it can produce a new A.A.
- It may or may not interfere with protein synthesis.

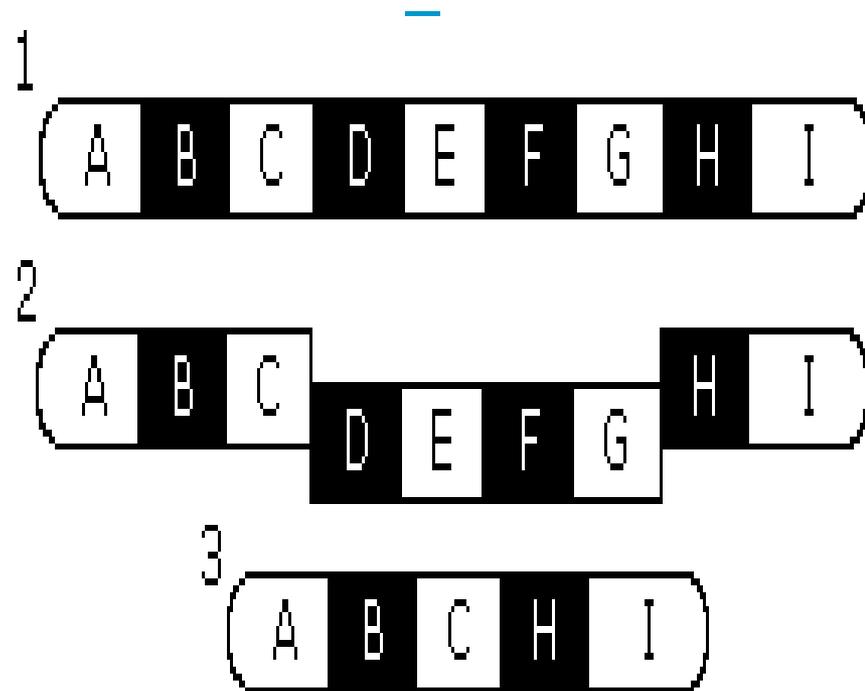


FRAMESHIFT MUTATION

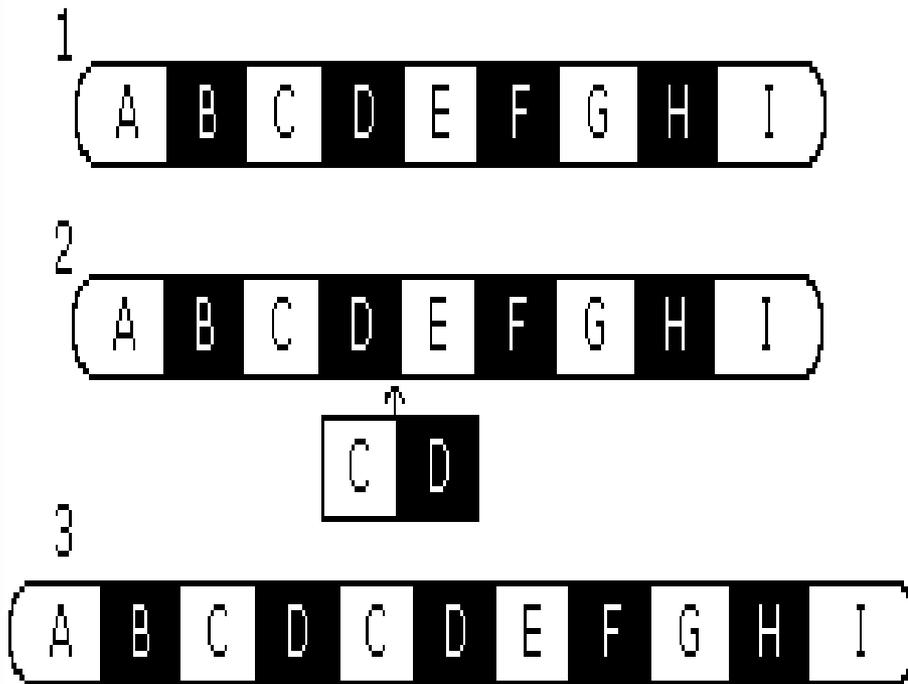
- A mutation in which a single base pair is added or deleted. This causes a shift in the reading of the codons by one base
- Insertion or Deletion one base of a DNA molecule will change nearly every amino acid in the protein.

DELETION

- **Deletion**- The loss of an inner chromosomal fragment- or when an end of a chromosome breaks off.



INSERTION (DUPLICATION)



| **Insertion**- The presence of a chromosome segment more than once in the same chromosome.

Frameshift Mutations: Phage T4 gene e (lysozyme)

Wildtype ... lys . ser . pro . ser . leu . asn . ala . . .
 ... AAA . **AGU** . CCA . UCA . CUU . AAU . GCU . . .

Altered reading frame

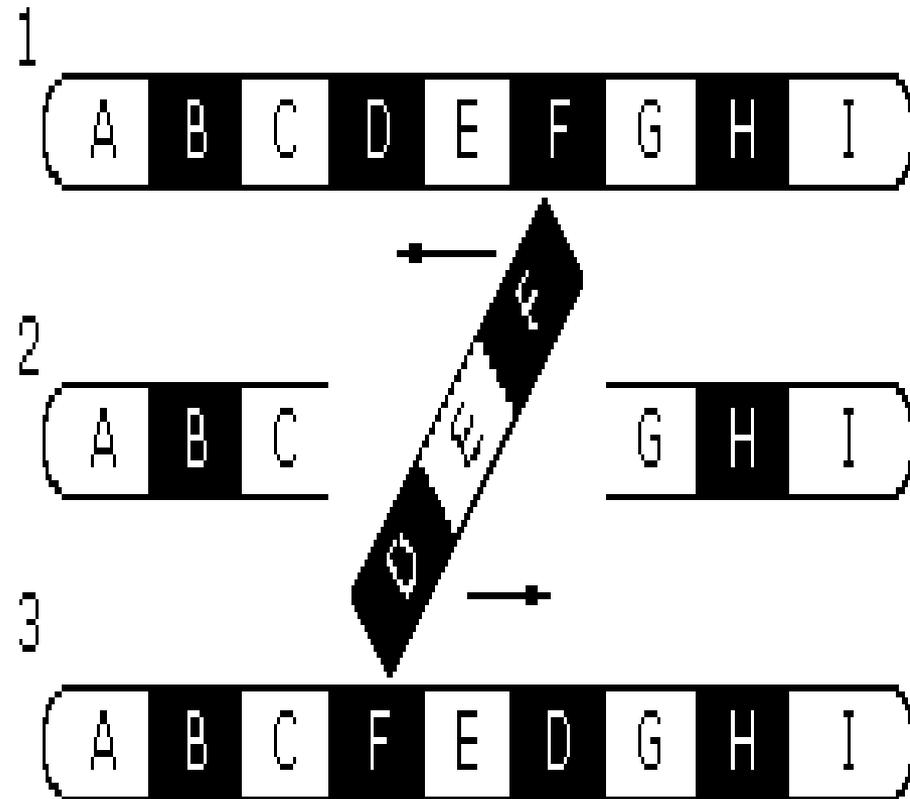
Deletion ... lys . val . his . his . leu . met . . .
(-) ... AAA . GUC . CAU . CAC . UUA . AUG . CU . . .

Altered reading frame

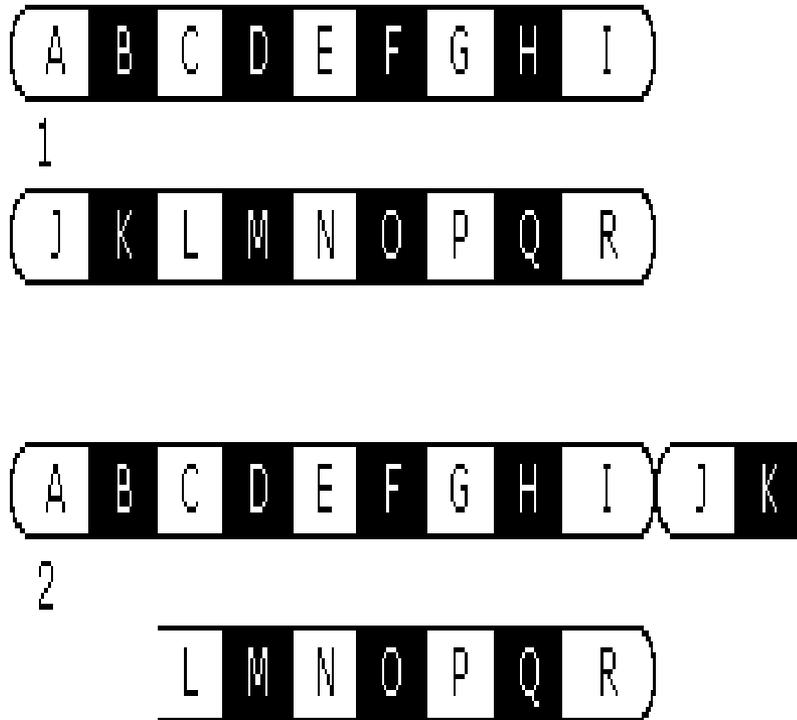
Insertion ... lys . ser . thr . ile . thr . stop
(+) ... AAA . AGU . **ACC** . AUC . ACU . UAA . UGC . U . . .

INVERSION

- **Inversion**- the change of direction of a chromosomal segment when it breaks out and is re-inserted backwards.



TRANSLOCATION

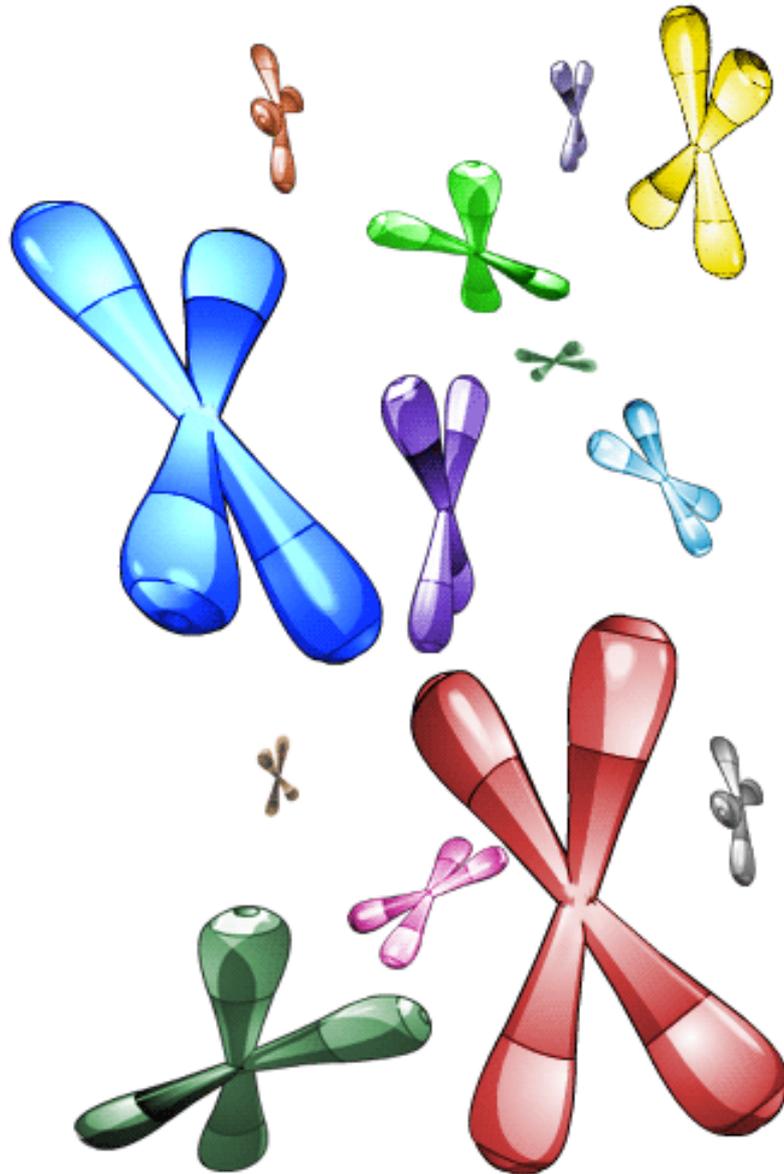


- **Translocation-**
The movement of chromosomal segments from one chromosome to another, non-homologous chromosome



Today's Assignment!!

- You are to draw, color, and label Chromosome Mutations on page 308.
- You may not trace!
- It must be on **PLAIN** white paper!!
- It needs to be as **large** as the paper...not too small!!
- Due tomorrow...so get busy!!





THANK YOU