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Genetic Mutations

- I. Structural Mutations in Chromosomes
- a. Point Mutations=mistake in a single nucleotide and only affects that specific codon
- i. Substitution
- Example:
- THE CAT ATE THE RAT
Changes ↓ to
THE RAT ATE THE RAT
- b. Frameshift Mutations=alters the nucleotide sequence from location of mutation and beyond in which all subsequent codons are altered.
- i. Insertion=inserts a nucleotide
- Example:
- THE CAT ATE THE RAT
Inserting a single letter
THE CRA TAT ETH ERA T
- ii. Deletion=removes a nucleotide
- Example:
- THE CAT ATE THE RAT
Removing a single letter
THE ATA TET HER AT
- iii. Duplication=specific type of insertion in which nucleotide is repeated
- Example:
- THE CAT ATE THE RAT
Duplicating nucleotide(s)
THE CAC_ATA TET HER A
- c. Inversion=nucleotides are inverted within the sequence
- Example:
- THE CAT ATE THE RAT
Inverts ↓ to
THE TAC ATE THE RAT
- d. Translocation=similar to crossing over BUT parts between *non-homologous* chromosomes are exchanged
- II. Aberrant Numbers of Chromosomes
- a. Aneuploidy=adding or removing entire chromosomes

- i. Monosomic=missing one chromosome
 - 1. Barr bodies=inactive X
 - 2. Turner syndrome or Monosomy X=sterile; secondary sex characteristics with estrogen replacement therapy; normal intelligence
 - ii. Trisomic=has an extra chromosome
 - 1. Down syndrome is the result of trisomy of chromosome 21
 - 2. Trisomy X is female with three X chromosomes
 - iii. Other disorders from aneuploidy in autosomes
 - iv. Other disorders from aneuploidy of sex chromosomes
 - 1. Klinefelter syndrome=XXY males; sterile; inactive X; some breast enlargement; normal intelligence
 - 2. XYY males normal, tend to be a little taller
- b. Polyploidy=adding or removing entire *sets* of chromosomes