Name:_____ **Hour:**_____

Mutations and Karyotypes

Mutation - a change in the structure or amount of genetic material (DNA)

- Mutations can occur naturally during the replication (cell cycle), during the divisions of meiosis, or due to environment (like exposure to radiation or chemicals)
- Mutations can be small and only affect single nucleotides all the way up to entire Chromosome can be altered, missing, or deleted
- Mutations have many possible effects
  - Silent - have no effect
  - Create variation - such as different expressions of a trait
  - Major consequences - causes disorders
- Only mutations that occur in gametes (sex cells) are passed onto the offspring
  - Could be a mutation that the parent also has and genetically has passed on because it is in the DNA of all their cells
  - Could be a mutation that happened during meiosis

Chromosomal Mutations

Non-disjunction = this type of mutation happens when a pair of homologous chromosomes or a pair of sister chromatids fail to seperate during one of the divisions of meiosis

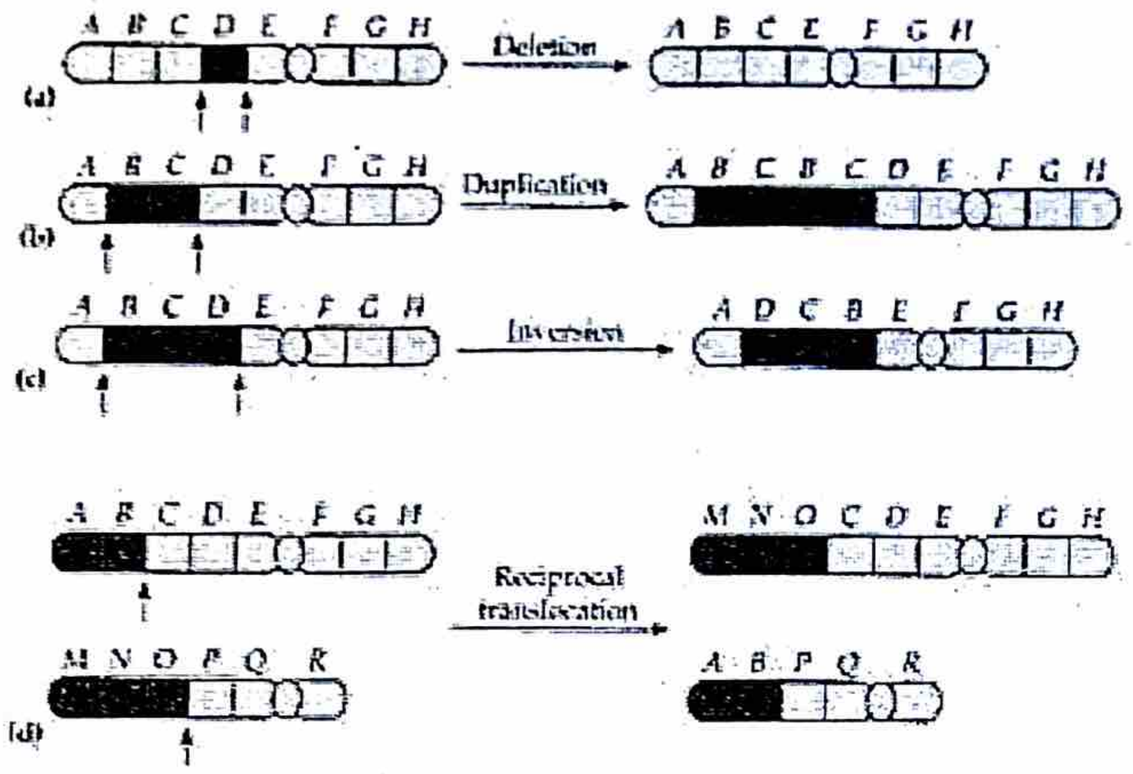
- Causes an additional chromosome to be passed onto the offspring

Deletion = a segment up to an entire chromosome is missing and has been destroyed or removed

Duplication = a segment of a chromosome is replicated twice causes a duplicate of genes

Inversion = a segment of a chromosome is rearranged and "flipped" backwards

Translocation = a segment of one chromosome is switched with another causing genes to land on chromosomes they dont belong with



How can we identify mutations?

- To look for chromosomal mutations we can analyze a person's Karyotype

Karyotype - is an organized profile of a person's chromosomes

- Organized by homologous pairs
  - Same size and same banding pattern allows scientists to identify pairs
- Chromosomes are arranged from largest to smallest, with their pairs, with the exception of sex chromosomes (they are listed last)
- Pairs are numbered from largest to smallest
- Females have 2 X chromosomes, males have an X and a Y (the Y is not the same size as the X)