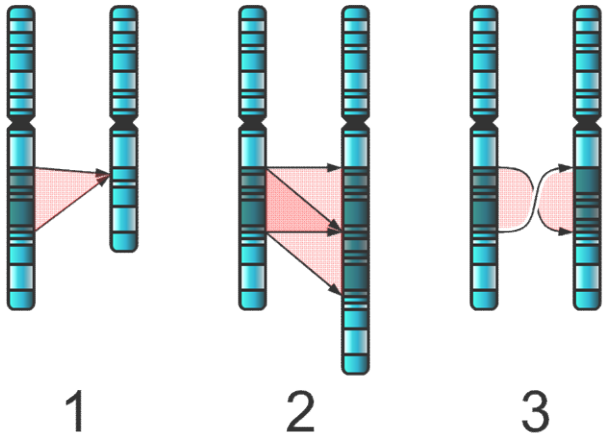
Mutations: Structural Chromosome Abnormalities

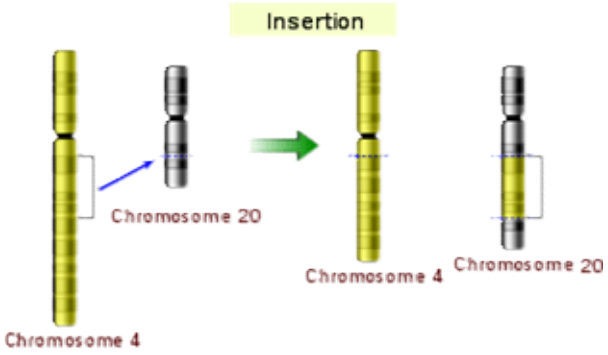
One-Chromosome Mutations

- Deletions: A portion of the chromosome is missing or deleted. Known disorders in humans include Wolf-Hirschhorn syndrome (born with numerous deformations) and Jacobsen syndrome (heart, facial, skeletal defects).
- Duplications: A portion of the chromosome is duplicated, resulting in extra genetic material. Known human disorders include Charcot-Marie-Tooth disease type 1A (muscle disorder).
- Inversions: A portion of the chromosome trades places with an adjacent portion of the same chromosome. Inversions typically do not cause abnormalities.

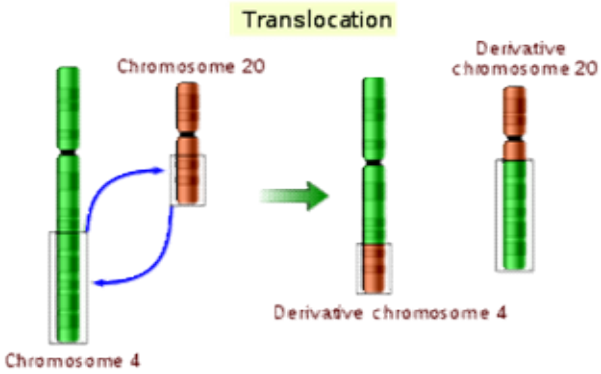


The three major single chromosome mutations; deletion (1), duplication (2) and inversion (3).

Two-Chromosome Mutations



- Insertions: One or more extra nucleotide is inserted into the DNA of another chromosome. Insertions can cause hemophilia, cancer, and muscular dystrophy.



- Translocations: Two chromosomes swap a portion of their DNA. Translocations can cause cancer, infertility, schizophrenia, and leukemia.