CONCEPT: GENOME EVOLUTION

Mutations in Evolution

● Accumulations in **mutations** (DNA changes) promote genome _____

□ **Point mutations** are changes in a single nucleotide pair

- Can be beneficial or detrimental to the organisms

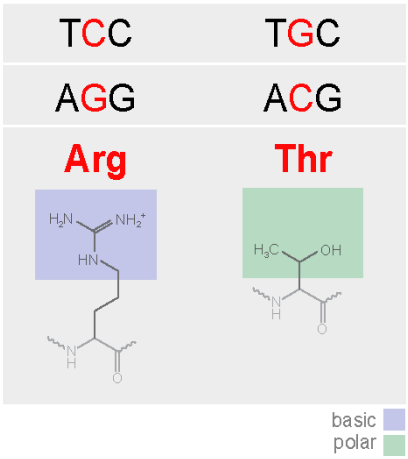
- Arise from errors in DNA replication

□ Mutations can occur in the gene, or in regulatory DNA

- Gene: effects activity, interaction, stability and is easy to spot

- Regulatory DNA: effects how genes are expressed and are not so easy to find

EXAMPLE: Point mutations can affect amino acid sequence



Gene Duplication in Evolution

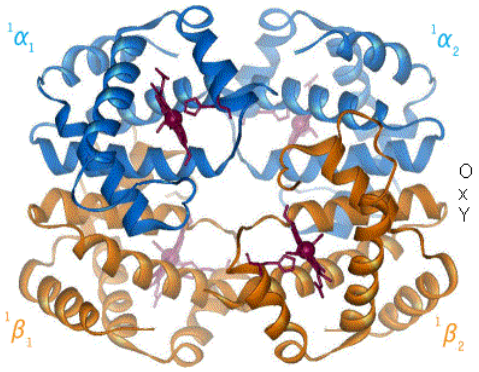
● **Gene duplication** is a main driver of genomic evolution

□ **Gene families** are groups of genes with similar sequences but _____ functions

- Once a gene is duplicated, each copy is free to accumulate mutations that result in different functions

- Gene families are the result of gene duplication

EXAMPLE: Structure of hemoglobin demonstrates gene families



□ Gene duplication arises from _____ *crossing over* during mitosis

- Misalignment of chromosomes during *homologous recombination* can lead to lopsided genetic exchange

- Result: one chromosome with extra gene copy and one with no copy

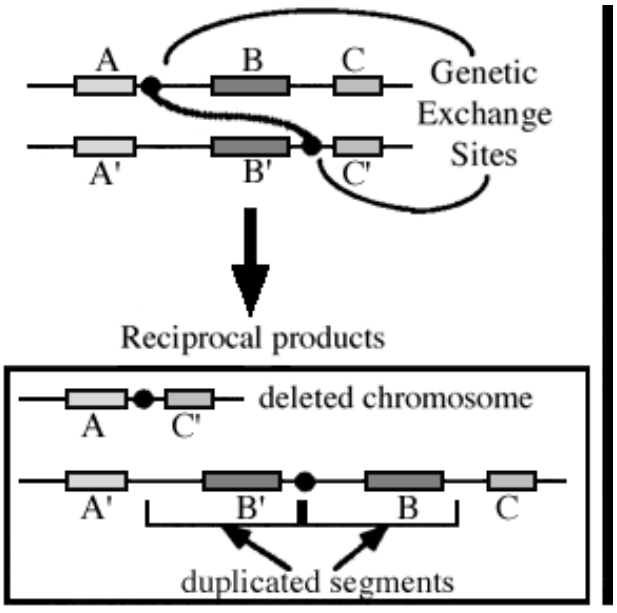
□ **Pseudogenes** are duplicated genes that have lost their functional ability but remain in the genome

- *Processed pseudogenes* occur by changing a mRNA to a DNA and integrating it into a chromosome

□ **Whole genome duplication** is when the entire genome of an organisms is copied and retained in a single cell

- **Polyploidization** (whole genome duplications) are common in fungi and plants

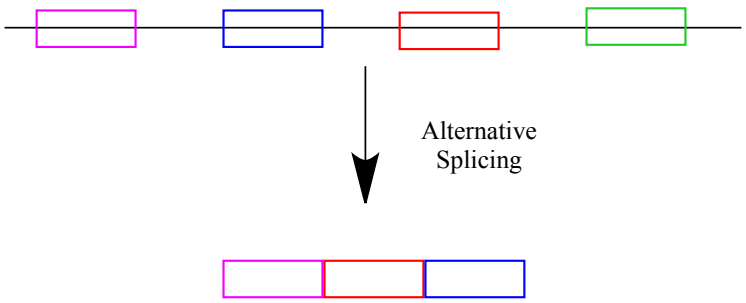
EXAMPLE: Unequal crossing over results in duplicated genes



Introns and Splicing in Evolution

- The presence of *introns* allows for gene shuffling and gene evolution
    - □ Genes of most organisms contain *Introns* and *Exons* (not completely universal ex: Histone proteins)
        - **Introns** are noncoding regions of a gene that are cut out during gene processing
        - **Exons** are the coding regions of a gene
    - □ **Alternative splicing** is the combining of exons from one gene in new orders (occurs in 50-90% of human genes)
        - Produce *isoforms* which are different forms of the same protein produced through alternative splicing
    - □ **Exon shuffling** is the combining of exons from two _____ genes
        - Can also occur if exons are duplicated or moved to different genomic location

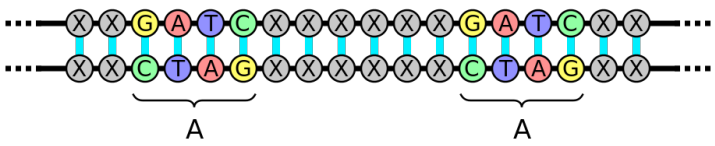
EXAMPLE: Model of alternative splicing



Repetitive DNA sequences in Evolution

- **Repetitive DNA sequences** are have evolved in the genomes and are extremely common
    - □ **Simple-sequence repeats** are arrays of thousands of copies of a short sequence (1-500 nucleotide)
        - *Drosophila:* ACAAACT
        - Not transcribed, and contain no genetic information
    - □ **Mobile genetic elements** are DNA sequences that can _____ through the genome
        - Contain repetitive DNA flanking protein coding regions
        - **Transposons** are mobile genetic elements and can move through RNA or DNA intermediates
        - Insert anywhere in a gene, and can effect gene structure or regulation

EXAMPLE: Example of sequence repeats in the genetic code



PRACTICE:

1. Which of the following is not a driver of genome evolution?
    a. Mutations
    b. Gene duplication
    c. Alternative Splicing
    d. Histone modifications

2. Which of the following genomic changes are most likely to cause pseudogenes?
    a. Point Mutations
    b. Gene Duplications
    c. Exon Shuffling
    d. Transposons

3. Protein isoforms are created through which process?
    a. Point Mutations
    b. Alternative Splicing
    c. Exon Shuffling
    d. Simple Sequence Repeats