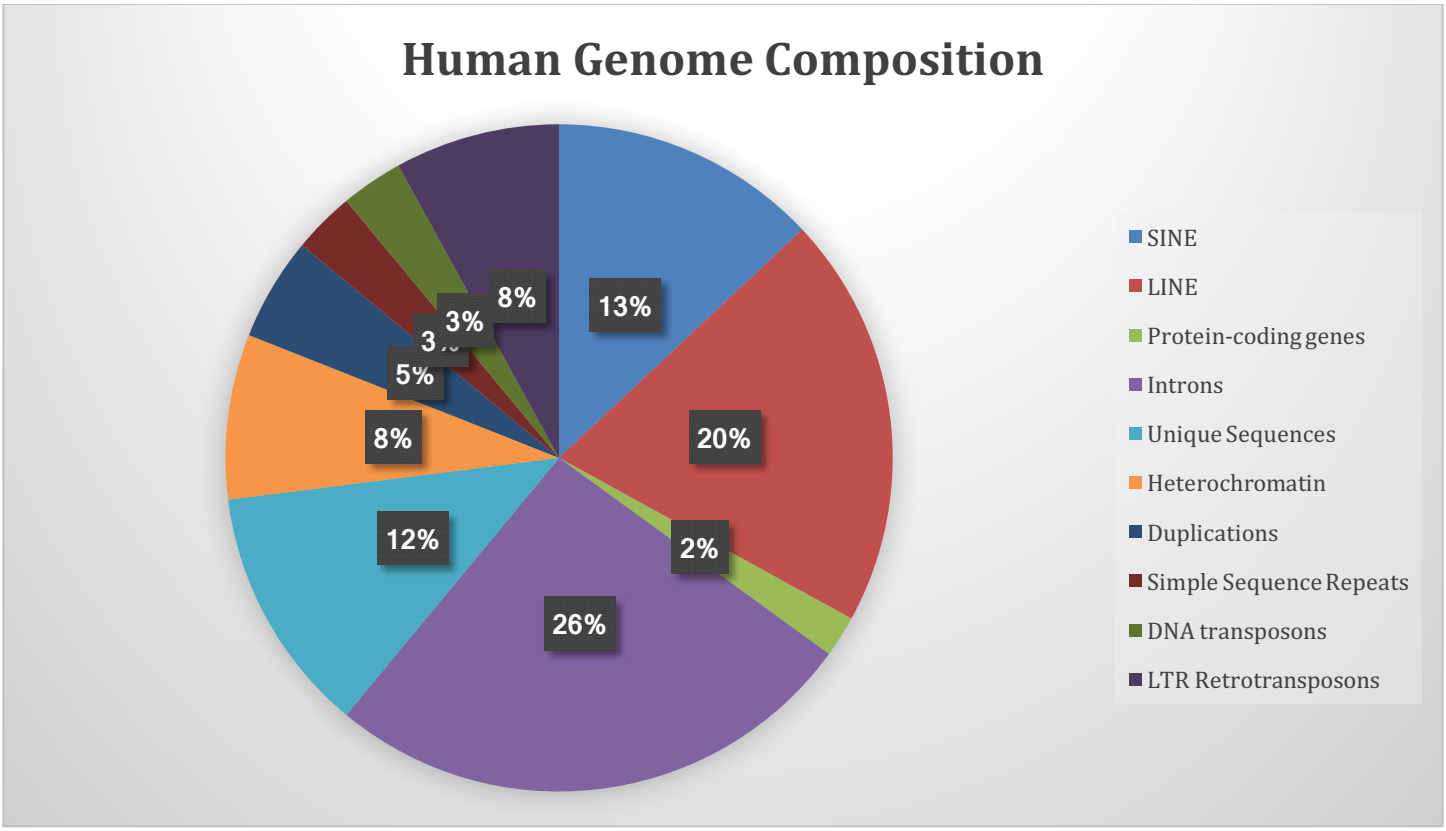
CONCEPT: HUMAN GENETIC VARIATION

- Sequencing of the human genome revealed knowledge about its structure, size, and composition
  - □ The human genome contains 3.2 x 10^9 nucleotide pairs organized in 23 sets of chromosomes
    - Less than 2% encode for proteins
  - □ _____ of the human genome is:
    - 20,000-25,000 protein coding genes (1.2% of genome)
    - 50% of *mobile genetic elements* or "jumping genes"
    - 9,000 functional RNAs
  - □ 5% of the human genome is highly conserved in other organisms

EXAMPLE: Human genome composition



● Comparison of the genomic sequence between humans and other organisms demonstrates similarities and differences

□ Prokaryotes were first sequenced in 1995

- 90% of genome is protein coding

□ Yeast were first sequenced in 1996

- 70% of genome is protein coding

□ *C. elegans* (worms) were first sequenced in 1998

- 25% of genome is protein coding

□ *Drosophila melanogaster* (fruit flies) was first sequenced in 2000

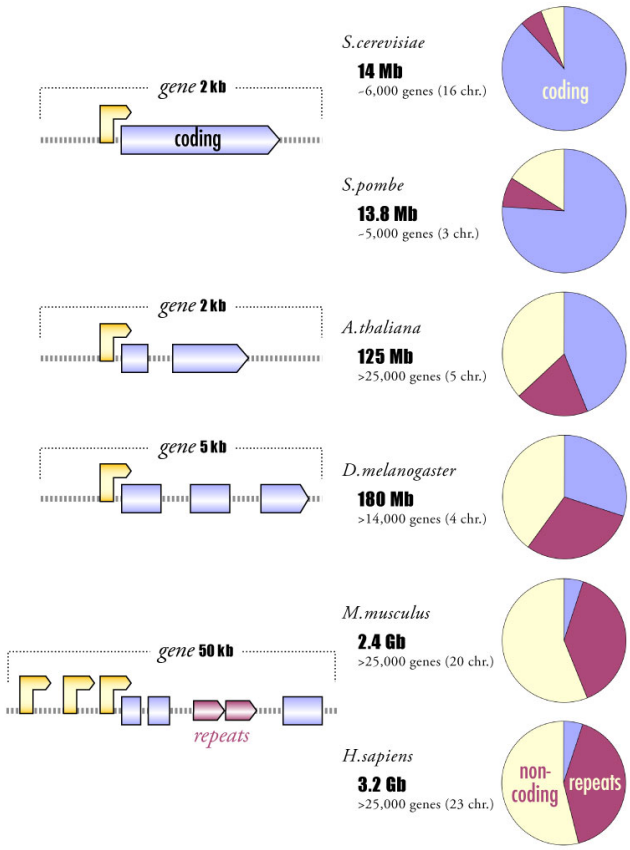
- 13% of genome is protein coding

□ *Arabidopsis* (flowering plant) were first sequenced in 2000

- 25% of genome is protein coding

□ Size of genome does not dictate complexity

EXAMPLE: Genomic comparisons of organisms



Human Evolution

● Chimpanzees and humans _____ from a common ancestor

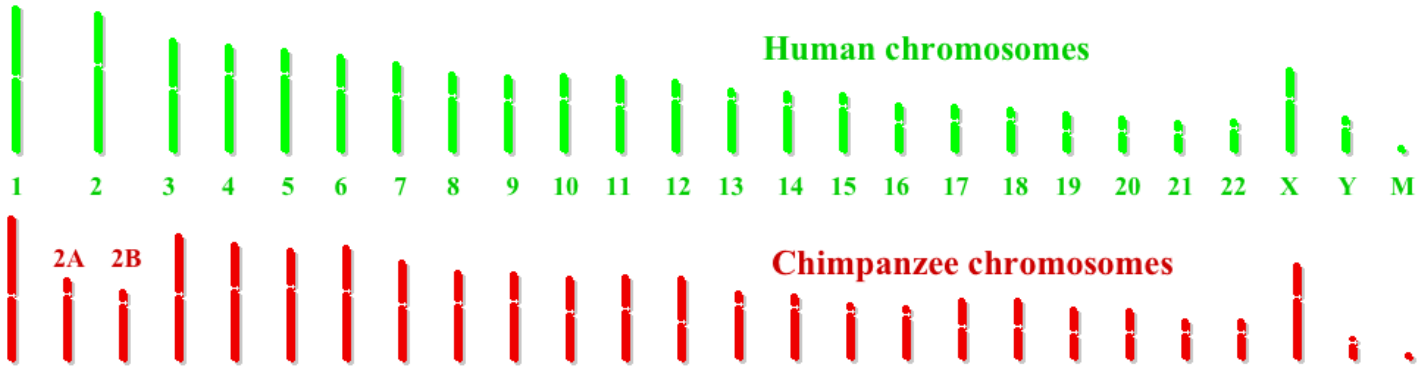
□ 98% similarity between the human and chimpanzee genomes

□ **Human accelerated regions** are conserved areas of genome where rapid evolution occurred in humans

- There are around 50 sites within the human genome

- 25% support changes near genes that control brain development

EXAMPLE: Comparison between human and chimpanzee chromosomes



● Human _____ exists between individuals

□ 1 in 1,000 nucleotides differs between one individual person and another

- 3 million genetic differences

□ **Single nucleotide polymorphisms (SNPs)** are differences in the genome of one population and another

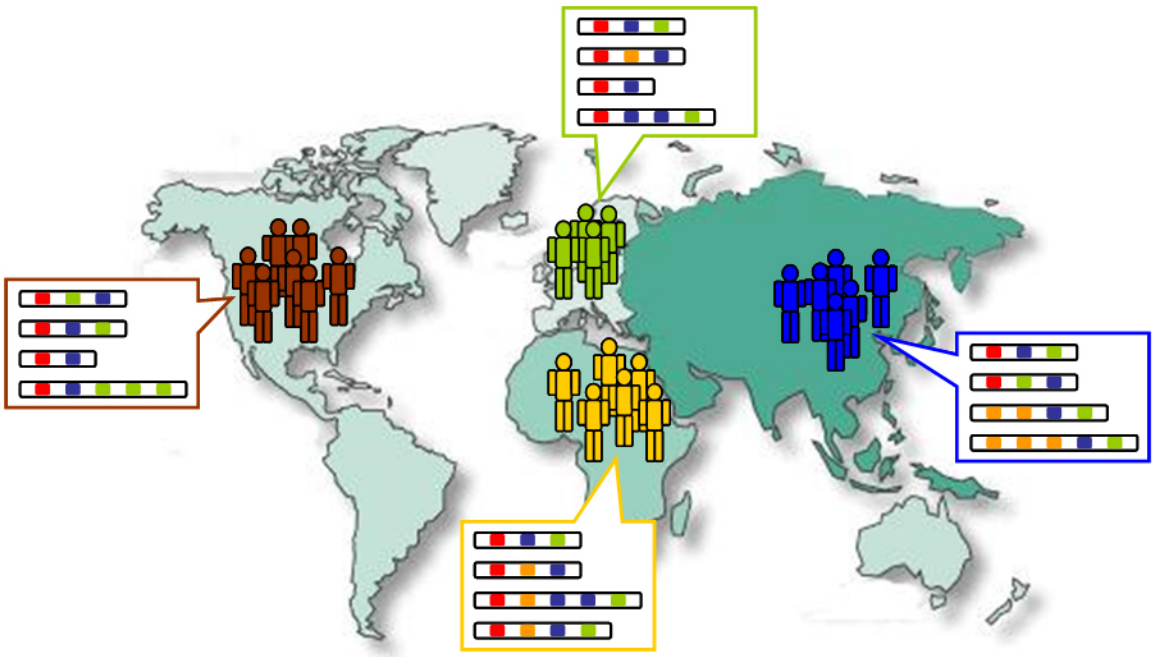
- Two randomly chosen people differ by 2.5 x 10^6 SNPs

□ **Copy-number variation** describes differing number of gene copies in one individual and another

□ **CA** repeats are strings of repeating C,A nucleotides and are very prone to mutations

- **DNA fingerprinting** uses these to identify specific individuals

EXAMPLE: Model of genetic variation in different human populations



PRACTICE

1. Which of the following is true regarding genomic genetic variations?
    a. Genomic size is proportional to genomic complexity
    b. Mobile genetic elements make up a very small proportion of the human genome
    c. Sequence variations between one individual and another occurs once every 1000 nucleotides
    d. CA repeats are extremely stable genetic elements found in the human genome

2. True or False: The majority of the human genome encodes for proteins.
    a. True
    b. False

3. Which of the following genomic variation refers to different number of gene copies between individuals and populations?
    a. Single nucleotide polymorphisms
    b. Copy number variants
    c. CA Repeats
    d. Transposons