CONCEPT: GENOMIC VARIATIONS

● There are _____ of different variations that exist between two individual's genomes

□ **Single nucleotide polymorphisms (SNPs)** are single nucleotide variations

- One in every 1000 bases is altered between two individuals (Around 18 million total SNPs in humans)

- Vast majority of SNPs are in silent regions of the genome

- SNPs are identified via multiple ways

- Southern blots of restriction enzyme cut DNA will result in different DNA lengths

- Other methods include PCR, and DNA microarrays

EXAMPLE: Example of a single nucleotide polymorphism

ACCTAGTTGCAATG ⟶ ACCTACTTGCAATG

□ **Deletion-insertion polymorphisms** (DIPs or In-Dels) are small deletions or insertions of genetic material

- Can _____- from 1-517 base pairs, and there are nearly 300,000 in the human genome

- One in every 10kb of DNA differs between two individual

□ **Simple sequence repeats** are 1,2, or 3 base sequences repeated 15-100 times

- A CA repeat is repeated once in every 30,000 base pairs in mammalian genomes

- Arise through DNA replication errors

- Can cause serious diseases like Huntington disease

EXAMPLE: Example of CA repeats

CACACACACACACACACACACACACACA

□ **Minisatellites** are repeats of 500pb to 20kb in size that are scattered throughout the genome

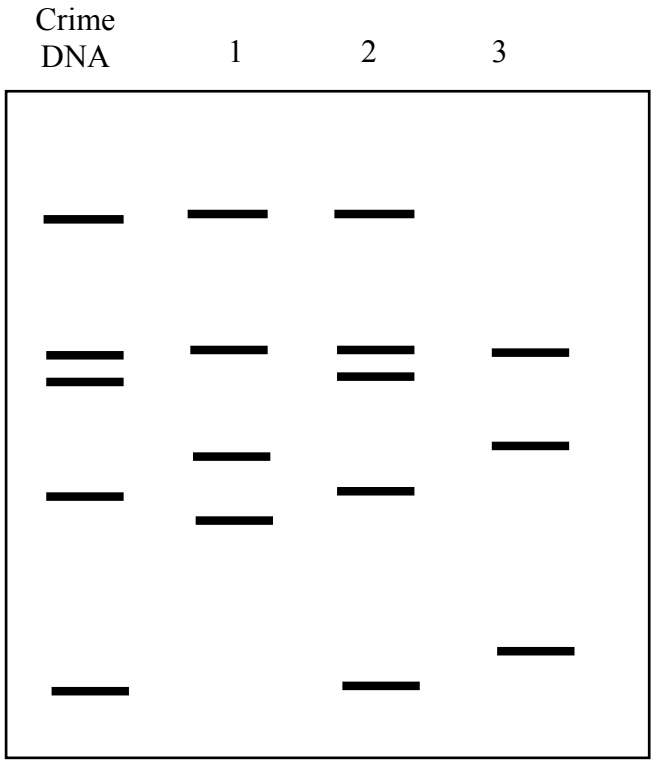
- **DNA fingerprint** is a pattern produced by detection of a genotype (minisatellites) at unlinked loci

- DNA fingerprinting pattern is used in crime investigations to identify a suspect's DNA

□ **Large scale deletions** can also differentiate human _____

- **Copy number variants** are polymorphism of up to 1mb in length

EXAMPLE: Example of a DNA Fingerprint



PRACTICE:

1. Which of the following is NOT an example of large genomic variations between two individual genomes?
    a. Single nucleotide polymorphisms
    b. Simple sequence repeats
    c. Dominant and Recessive alleles
    d. Minisatellites

2. Which of the following genomic variations can be detected through DNA fingerprinting?
    a. SNPs
    b. Simple sequence repeats
    c. Large scale deletions
    d. Minisatellites

3. Which of the following genomic variations is most common in human genomes?
    a. SNPs
    b. Simple sequence repeats
    c. Large scale deletions
    d. Minisatellites