Genetic Variation and Polymorphisms

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Course: Genetics II (MA 206)

Summer Class

Lecture-5

Date 23-08-2026

Outline

- What is Genetic Variation
- Sources of Genetic Variation.
- Single Nucleotide Polymorphisms
- Copy Number Variations
- Applications of SNPs and CNVs

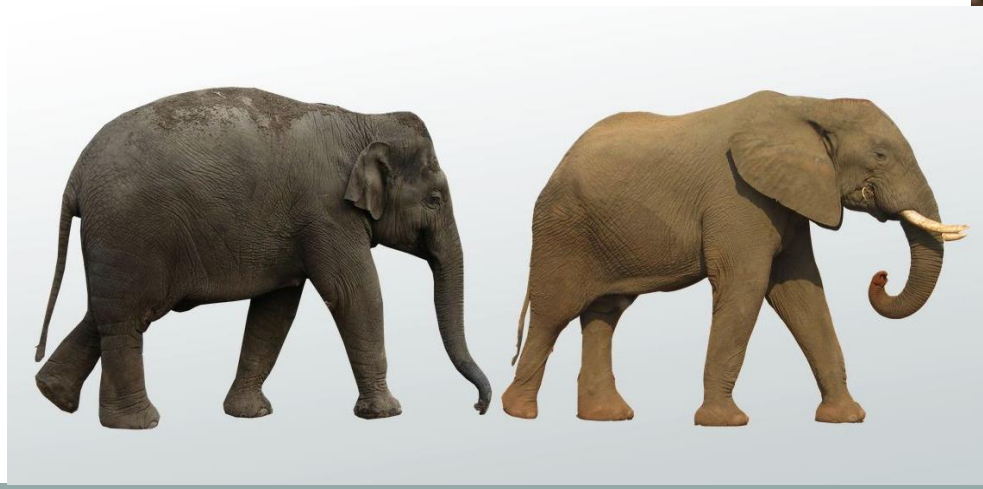
Objectives

- To understand and explain the fundamental principles of Genetic Variation and Polymorphisms.

❑ What is Genetic Variation?

- Genetic variation refers to **differences in the genetic makeup** among individuals within a **population or species**.

- It is the **foundation of evolution** and contributes to the **diversity of life**.



❖ Sources of Genetic Variation:

i. **Mutations**: Changes in the DNA sequence

ii. **Recombination**: Exchange of genetic material during meiosis.

iii. **Gene Flow**: Movement of genes between populations through migration.

iv. **Sexual Reproduction**: Combines genetic material from two parents, creating unique offspring.

❑ Importance of Genetic Variation

- Evolution and Natural Selection.
- Disease Resistance and Health
- Agriculture and Food Security
- Scientific and Medical Advances

❑ Polymorphisms:

❖ A polymorphism is a **genetic variation that occurs in at least 1% of the population.**

❖ It is a stable and heritable change.

❑ Types of Genetic Variation:

1. Single Nucleotide Polymorphisms (SNPs)
2. Insertions and Deletions (Indels)
3. **Copy Number Variations** (CNVs)
4. Structural Variations
5. Variable Number Tandem Repeats (VNTRs)

❑ Single Nucleotide Polymorphisms :

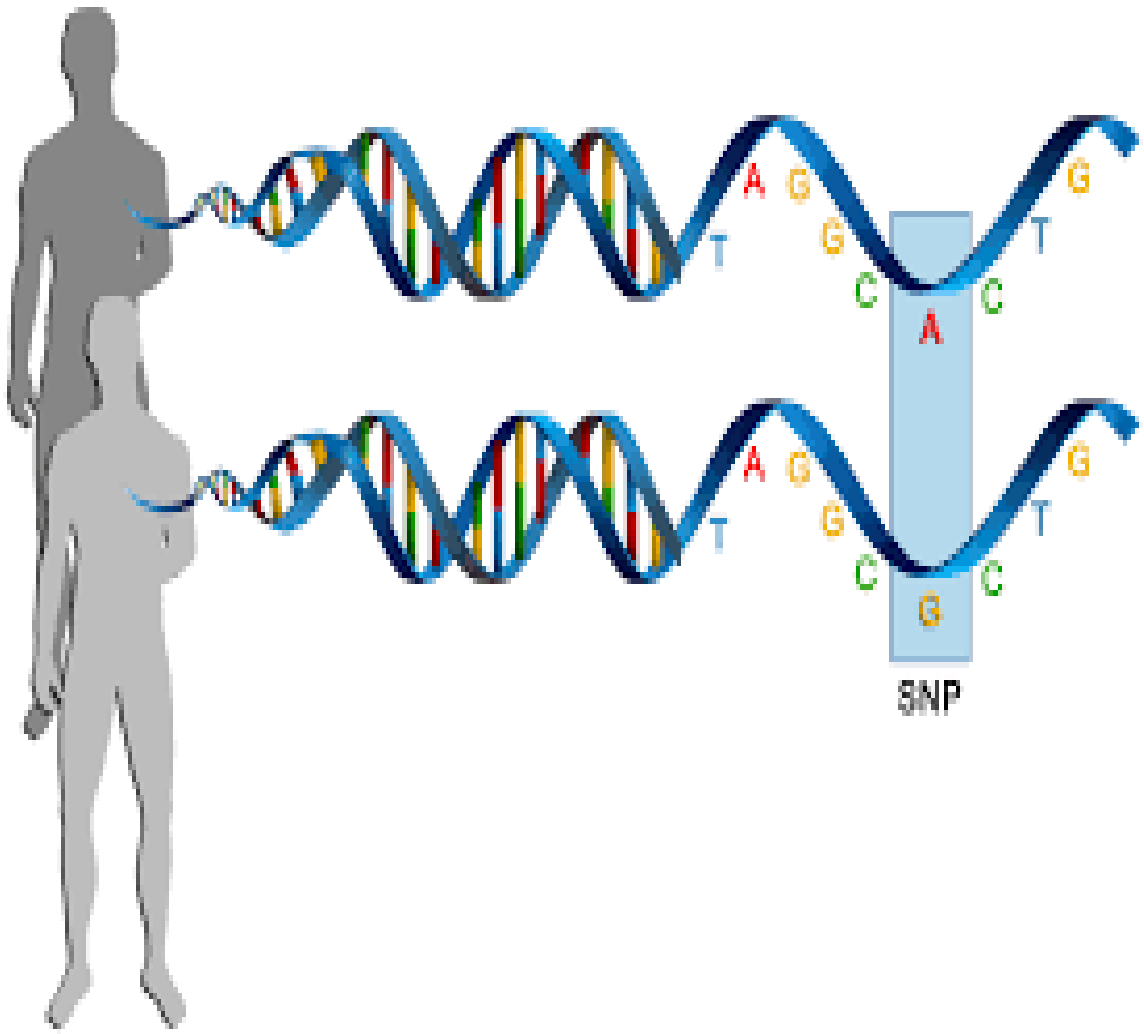
Definition: A SNP is a **variation at a single position** in a DNA sequence among individuals.

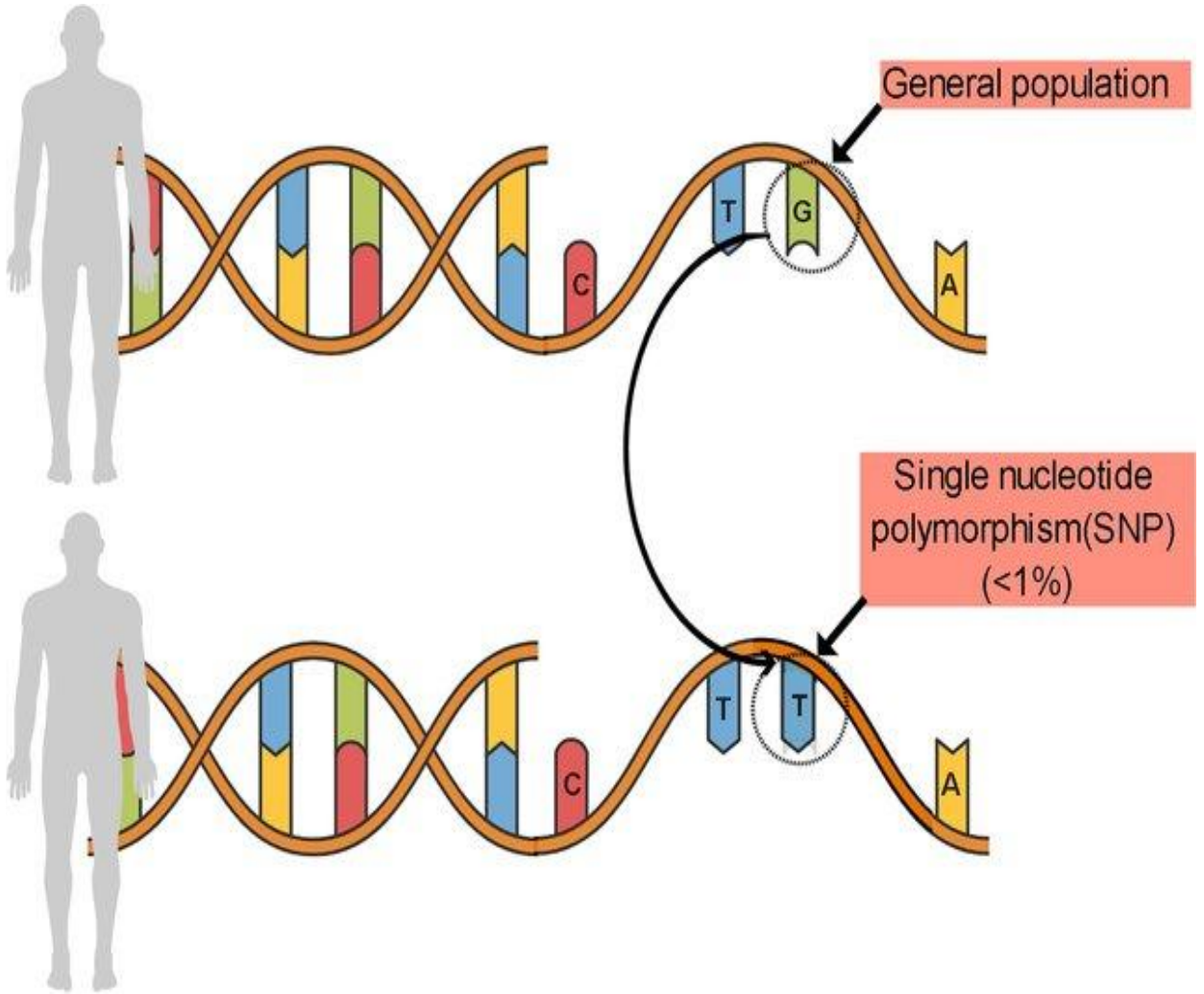
Must occur in **at least 1% of the population** to be considered a polymorphism

Example: If the DNA sequence is **AAGCCTA**, an SNP might change it to **AAGCTTA**.

SNPs occur roughly every **100-300 base pairs** in the human genome.

❑ Single Nucleotide Polymorphisms :





❑ Characteristics of SNPs

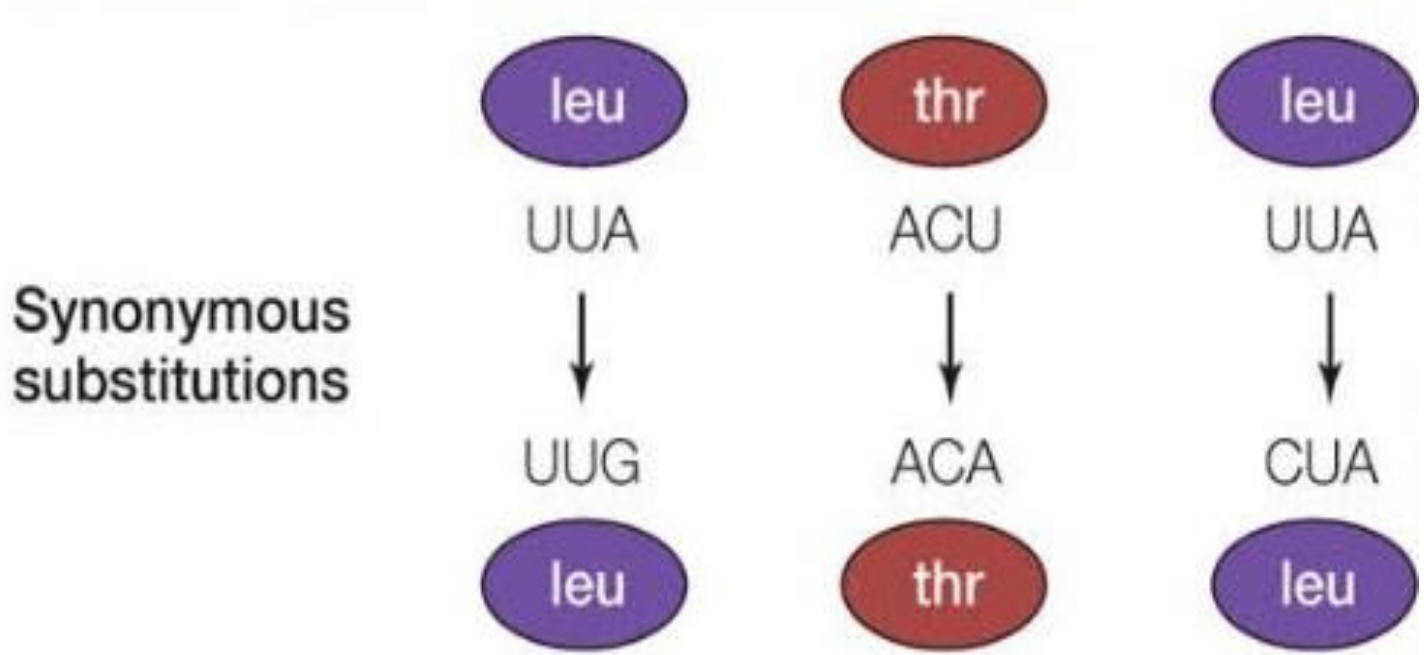
1. Most common type of genetic variation.

2. Can occur in coding or non-coding regions of the genome.

3. May or may not have a functional effect.

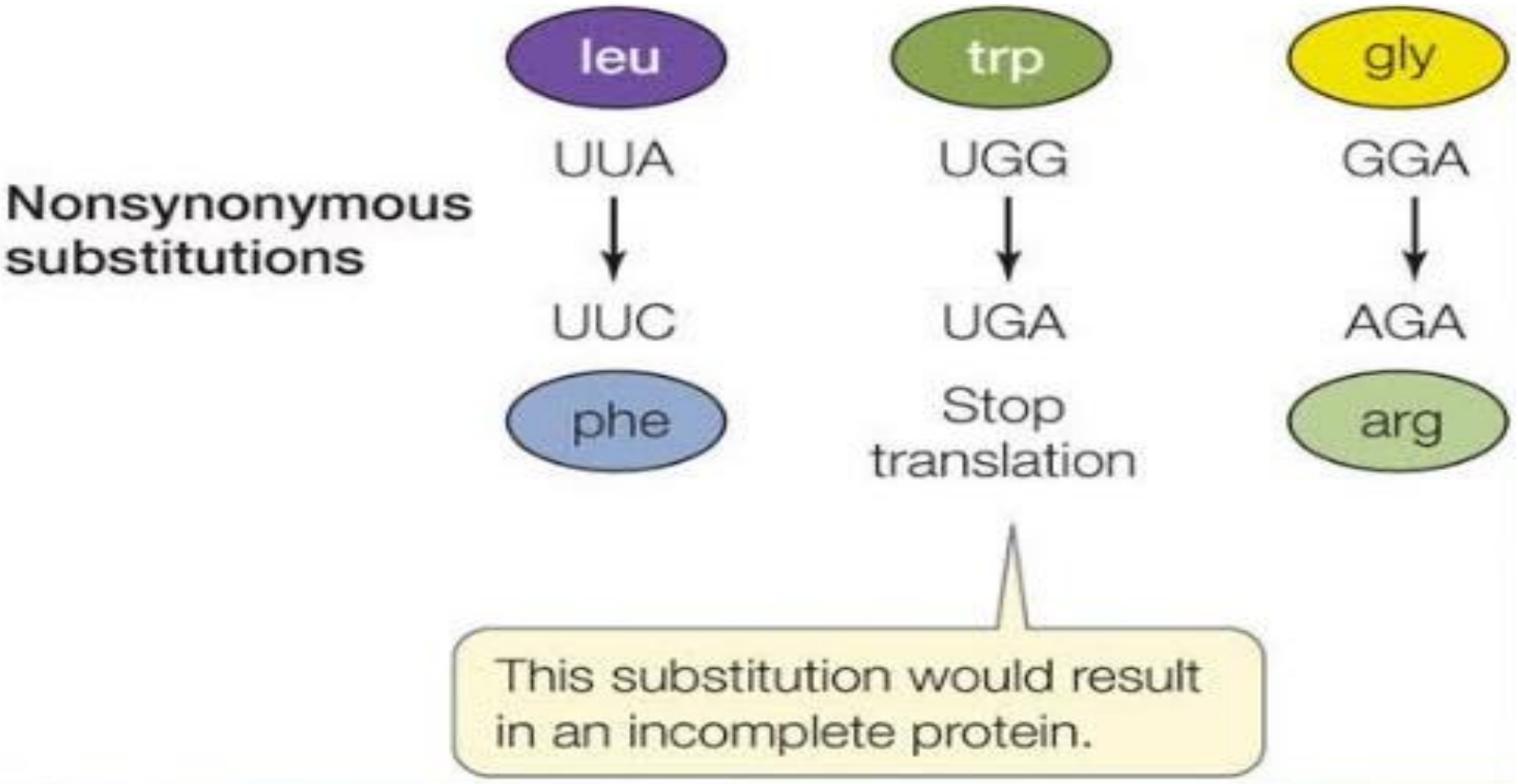
❑ Types of SNPs:

1. **Synonymous SNPs**: Do not change the amino acid sequence of the protein (**silent mutations**).



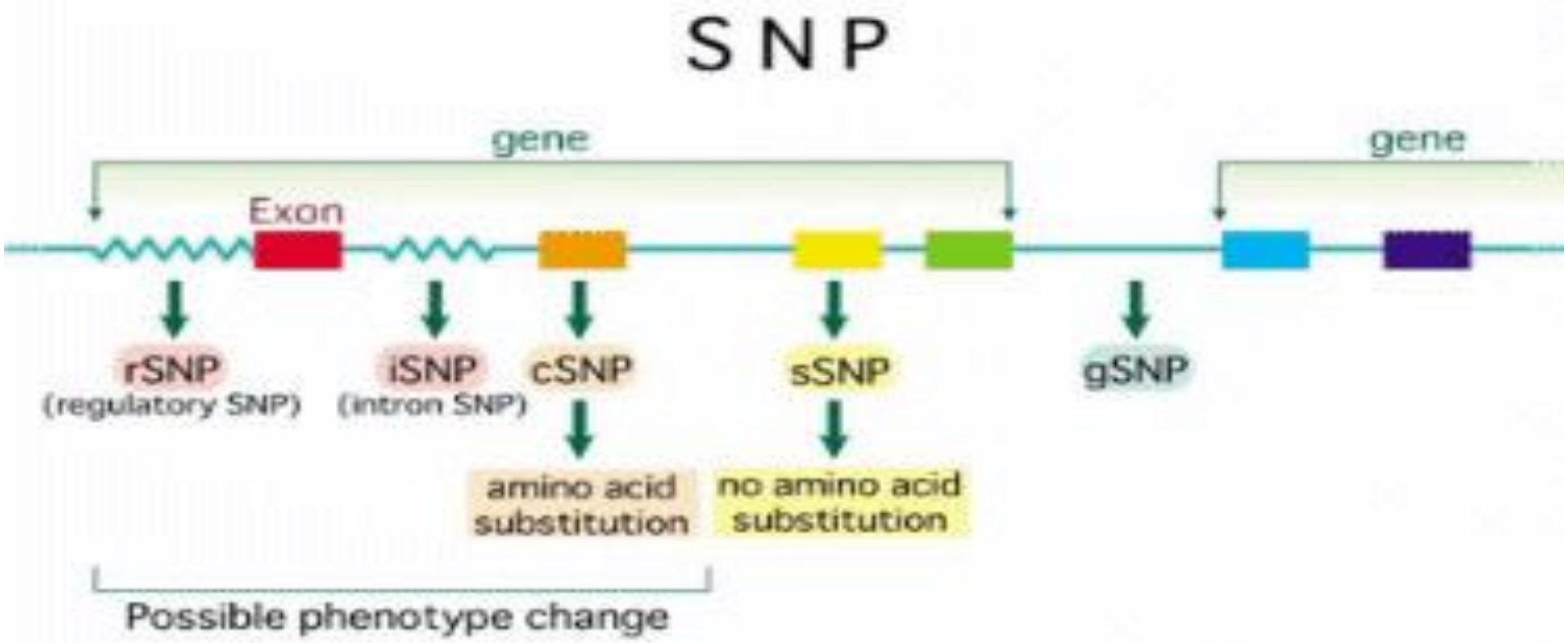
2. **Non-synonymous SNPs:**

- ✓ Change the amino acid sequence (**missense or nonsense mutations**).



3. **Regulatory SNPs**:

- ✓ Occur in regulatory regions (e.g., promoters, enhancers) and affect gene expression.



❑ Importance of SNPs

1. Genetic Markers:

- ✓ Used in genome-wide association studies (GWAS) to identify disease-associated genes.

2. Pharmacogenomics:

- ✓ Influence drug metabolism and response (e.g., warfarin sensitivity).

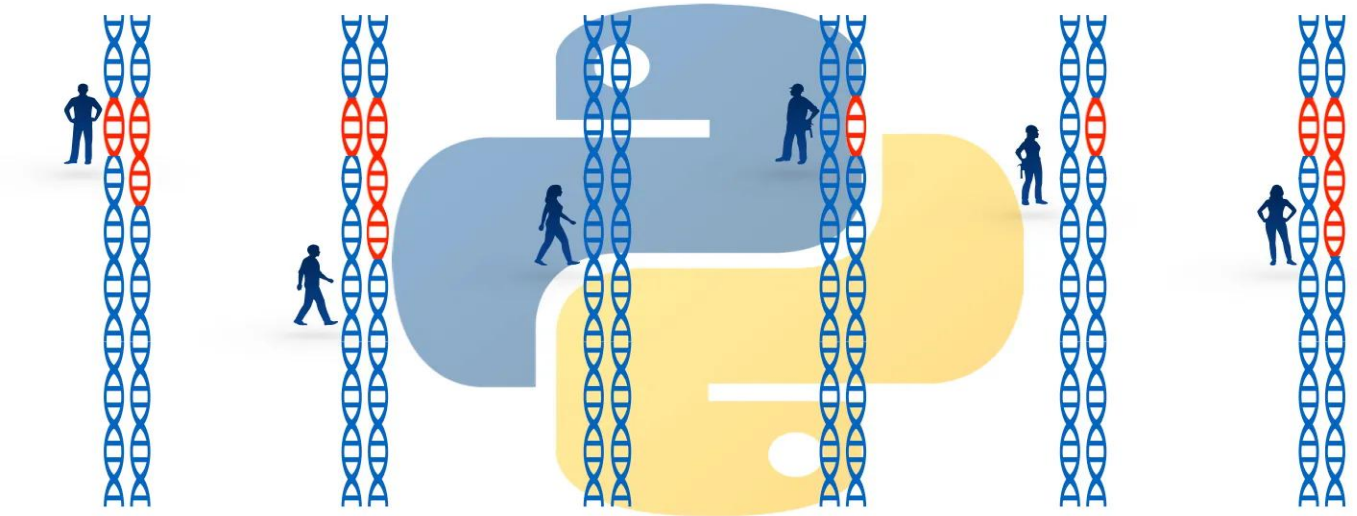
3. Disease Association:

- ✓ Linked to conditions like cancer, diabetes, and cardiovascular diseases.

❑ Copy Number Variations (CNVs):

❖ CNVs are structural variations involving changes in the number of copies of a particular DNA segment.

❖ Segments can range from 1 kilobase (kb) to several megabases (Mb) in size.



❑ Characteristics of CNVs:

- ❖ Can involve duplications, deletions, or complex rearrangements.
- ❖ May affect gene dosage, regulation, or function.
- ❖ Can be inherited.

❑ Types of CNVs

A. Deletions:

- ✓ Loss of a segment of DNA.

B. Duplications:

- ✓ Extra copies of a DNA segment.

C. Insertions:

- ✓ Addition of a DNA segment in a new location.

❑ Applications of SNPs and CNVs:

1. Medical Genetics:

❖ Identifying genetic risk factors for diseases .

2. Pharmacogenomics:

❖ SNPs can predict drug response and adverse effects.

3. Forensics:

❖ SNPs and CNVs used for DNA profiling and identification

4. Evolutionary Biology: Studying genetic diversity and population history

References

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3. Vafai SB, Mootha VK. "Mitochondrial disorders as windows into an ancient organelle." Nature, 2012.

Thanks