RAMA UNIVERSITY

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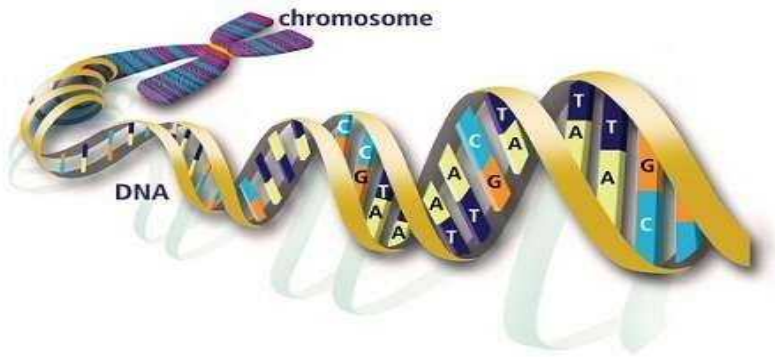
FACULTY OF ENGINEERING &TECHNOLOGY
DEPARTMENT OF BIOTECHNOLOGY

SINGLE NUCLEOTIDE POLYMORPHISM

- Single nucleotide polymorphisms or SNP (pronounced “snips”), are the most common type of genetic variation among peoples.

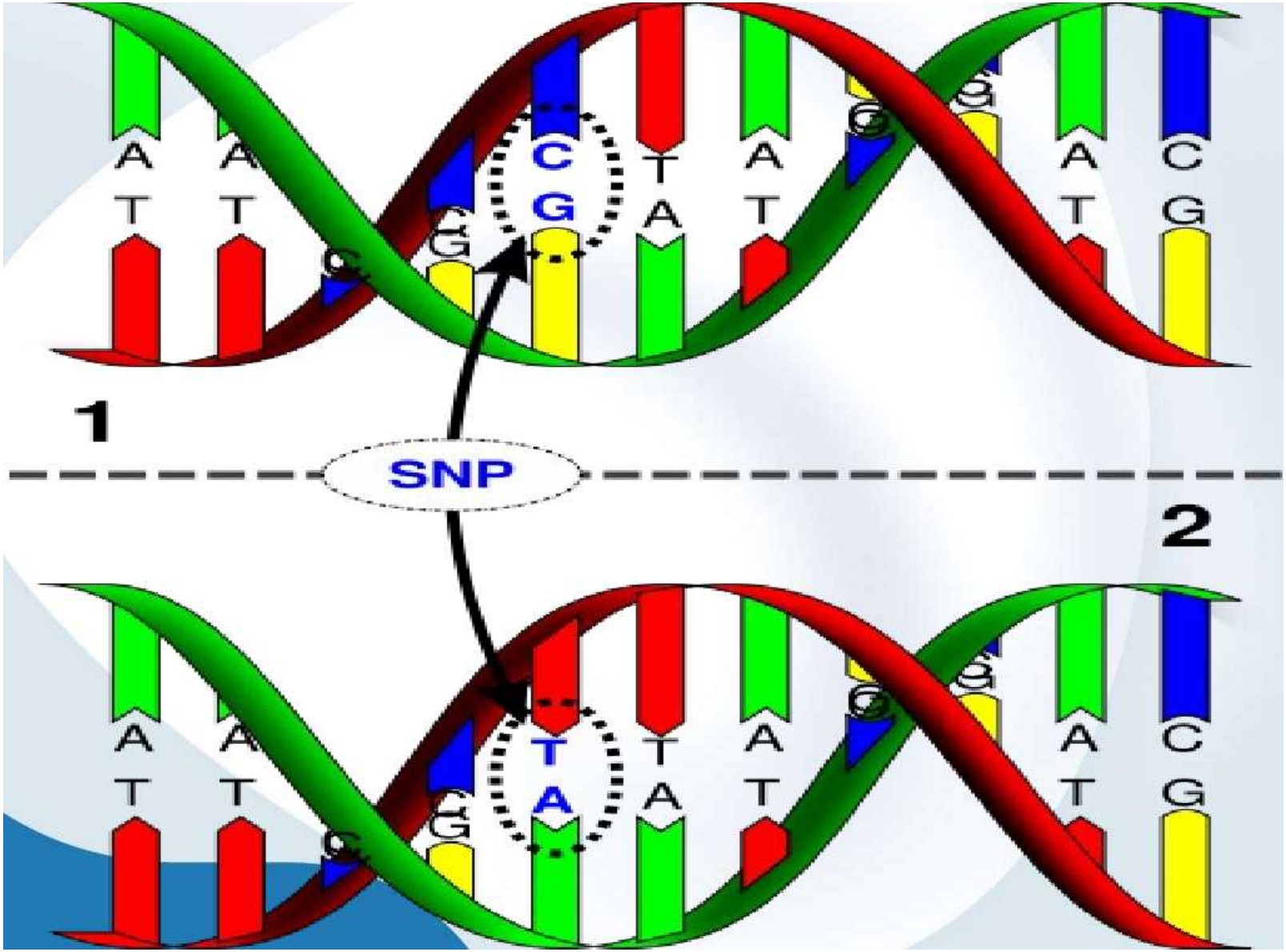
- Each SNP represents a difference in a single DNA building block, called a nucleotide



POLYMORPHISM

- Polymorphism is a generic term that means 'many shapes‘
- It is the ability to appear in different form

POLYMORPHISM



- It is a DNA sequence variation occurring when a single nucleotide A, T, C, or G in the genome differs between members of a species
- For example, two sequenced DNA fragments from different individuals, AAGC**C**TA to AAGC**T**TA, contain a difference in a single nucleotide

Some Facts

- In human beings, 99.9 percent bases are same. Remaining 0.1 percent makes a person unique.
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  - Different attributes / characteristics / traits
    - how a person looks,
    - diseases he or she develops.

- These variations can be:
  - Harmless (change in phenotype)
  - Harmful (diabetes, cancer, heart disease, Huntington's disease, and hemophilia)
  - Latent (variations found in coding and regulatory regions, are not harmful on their own, and the change in each gene only becomes apparent under certain conditions e.g. susceptibility to lung cancer)

SNPs facts

- SNPs are found in
  - coding and (mostly) non coding regions.
- Occur with a very high frequency
  - about 1 in 1000 bases to 1 in 100 to 300 bases.
- The abundance of SNPs and the ease with which they can be measured make these genetic variations significant.
- SNPs close to particular gene acts as a marker for that gene.
- SNPs in coding regions may alter the protein structure made by that coding region.

SNP MAPPING

- Sequence genomes of a large number of people
- Compare the base sequences to discover SNPs.
- Generate a single map of the human genome containing all possible SNPs

TYPES OF SNP

- Following are the types of SNP
- Non-coding region
- Coding region
  - Synonymous
  - Non synonymous
    - Missense
    - Nonsense

NON-CODING REGION

A segment of DNA that does comprise a gene and thus does not code for a protein .

CODING REGION

Regions of DNA/RNA sequences that code for proteins

Synonymous

A SNP in which both forms lead to the same polypeptide sequence is termed *synonymous* (sometimes called a silent mutation).

Non synonymous

If a different polypeptide sequence is produced they are non synonymous . A non synonymous change may either be missense or nonsense, where a missense change results in a different amino acid, while a nonsense change results in a premature stop codon.