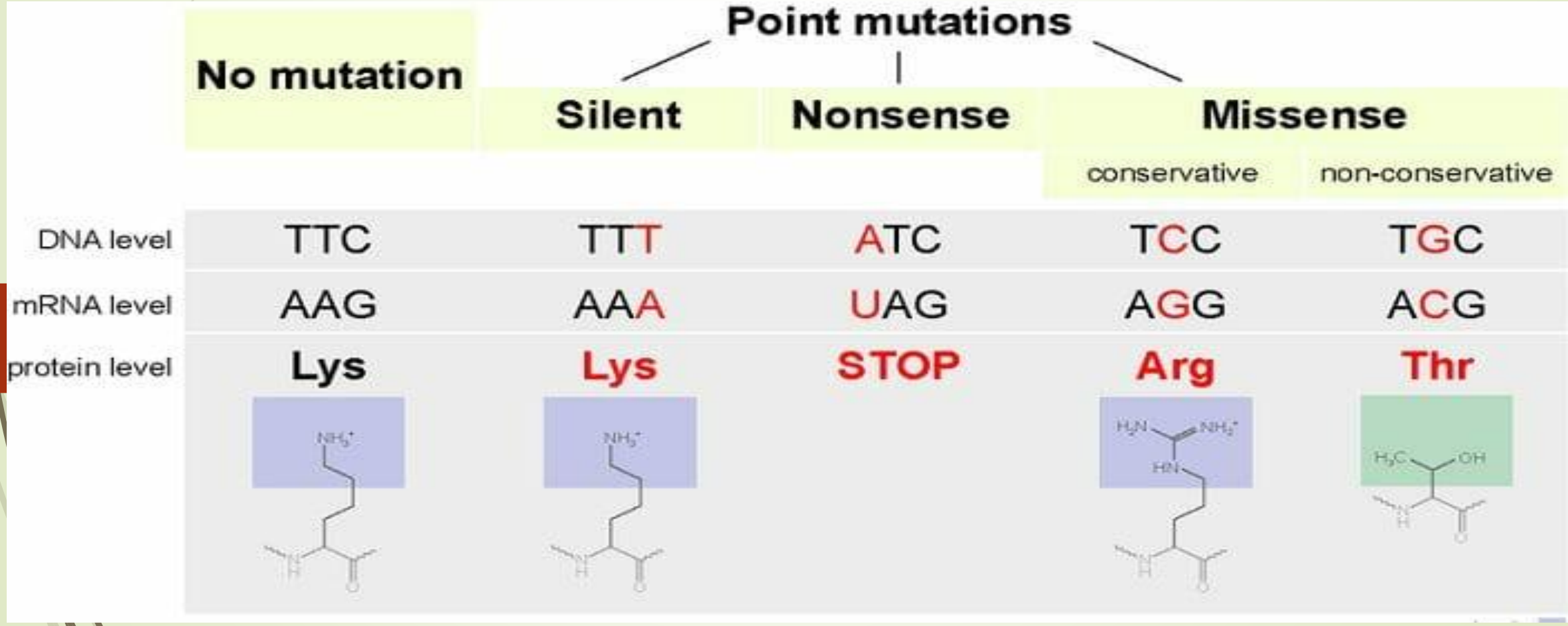
# MUTATIONS AND SINGLE NUCLEOTIDE POLYMORPHYSIMS (SNPs)

# MUTATION

- DNA mutations are heritable changes in DNA that, while often neutral or with subtle effects, can sometimes lead to observable phenotypic changes, potentially causing diseases or adaptations (K & E, 2017).
- Mutations are changes in the DNA sequence that can occur spontaneously or be induced by environmental factors.
- These can involve single base changes (point mutations), insertions, deletions, or larger rearrangement (Brown, 2002).

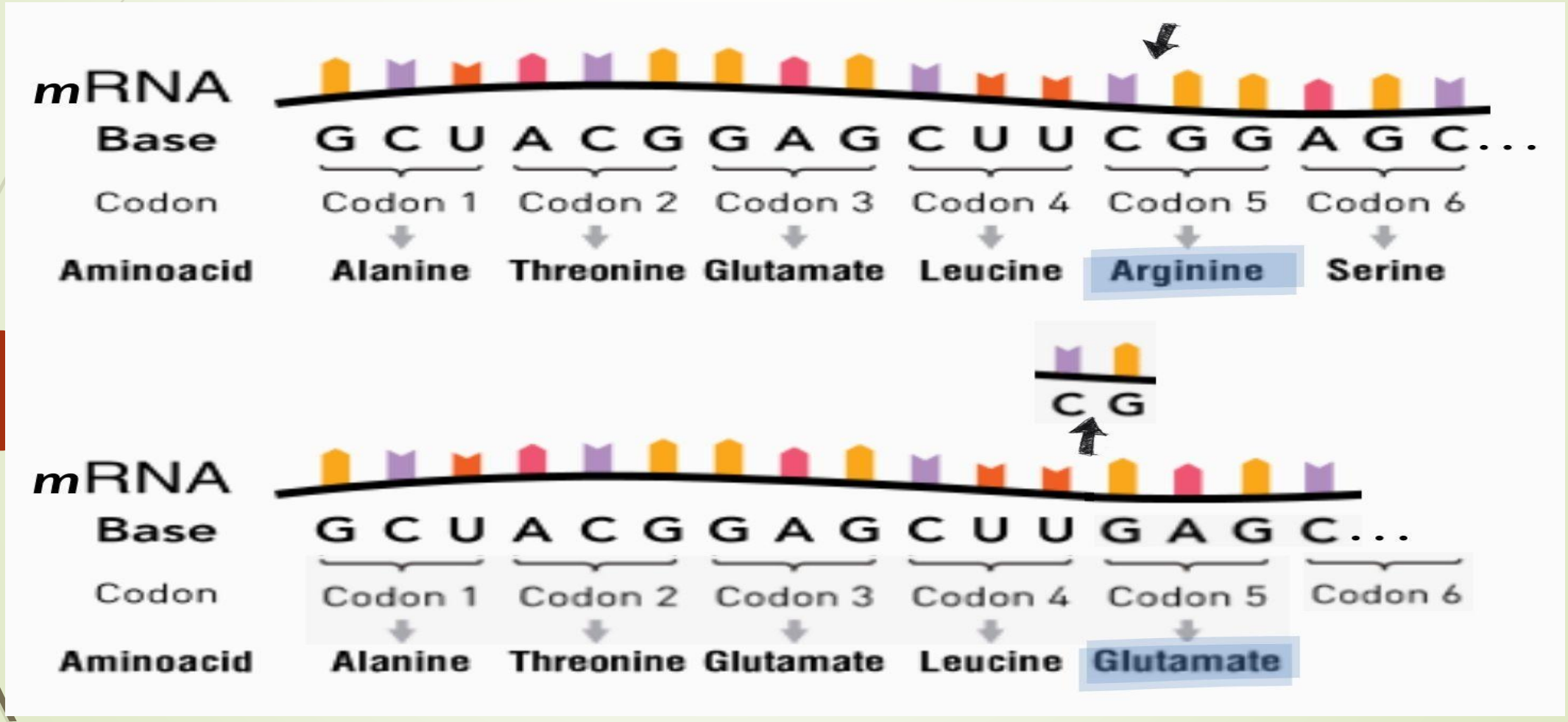
# Types of Mutations

- **Point mutation**: A single nucleotide change, such as a substitution, insertion, or deletion (Editors, 2016).



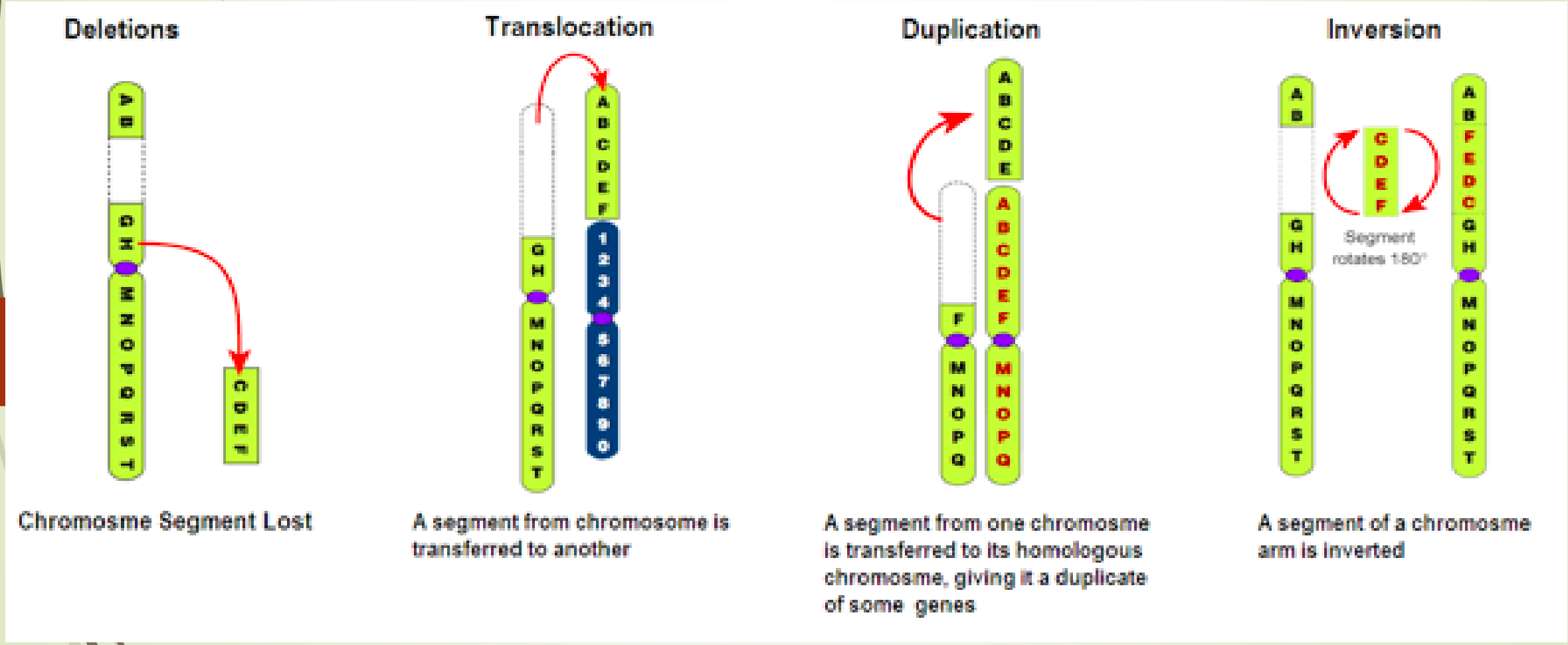
# Types of Mutations

- **Frameshift mutation**: A insertion or deletion of nucleotides that changes the reading frame of the genetic code (Kanakan et al., 2022).



# Types of Mutations

- **Chromosomal mutation**: A change in the number or structure of chromosomes (Loewe & Hill, 2010) .



# Factors Influencing Mutation

- **Genetic Factors**

- Genetic predisposition: Some individuals may be more prone to mutations due to their genetic makeup.
- Family history: A family history of genetic disorders or cancer can increase the risk of mutation (Gillio et al., 2007)..

- **Environmental Factors**

- Radiation exposure: Exposure to ionizing radiation, such as X-rays or gamma rays, can increase the risk of mutation.
- Chemical mutagens: Exposure to certain chemicals, such as pesticides or heavy metals, can increase the risk of mutation.

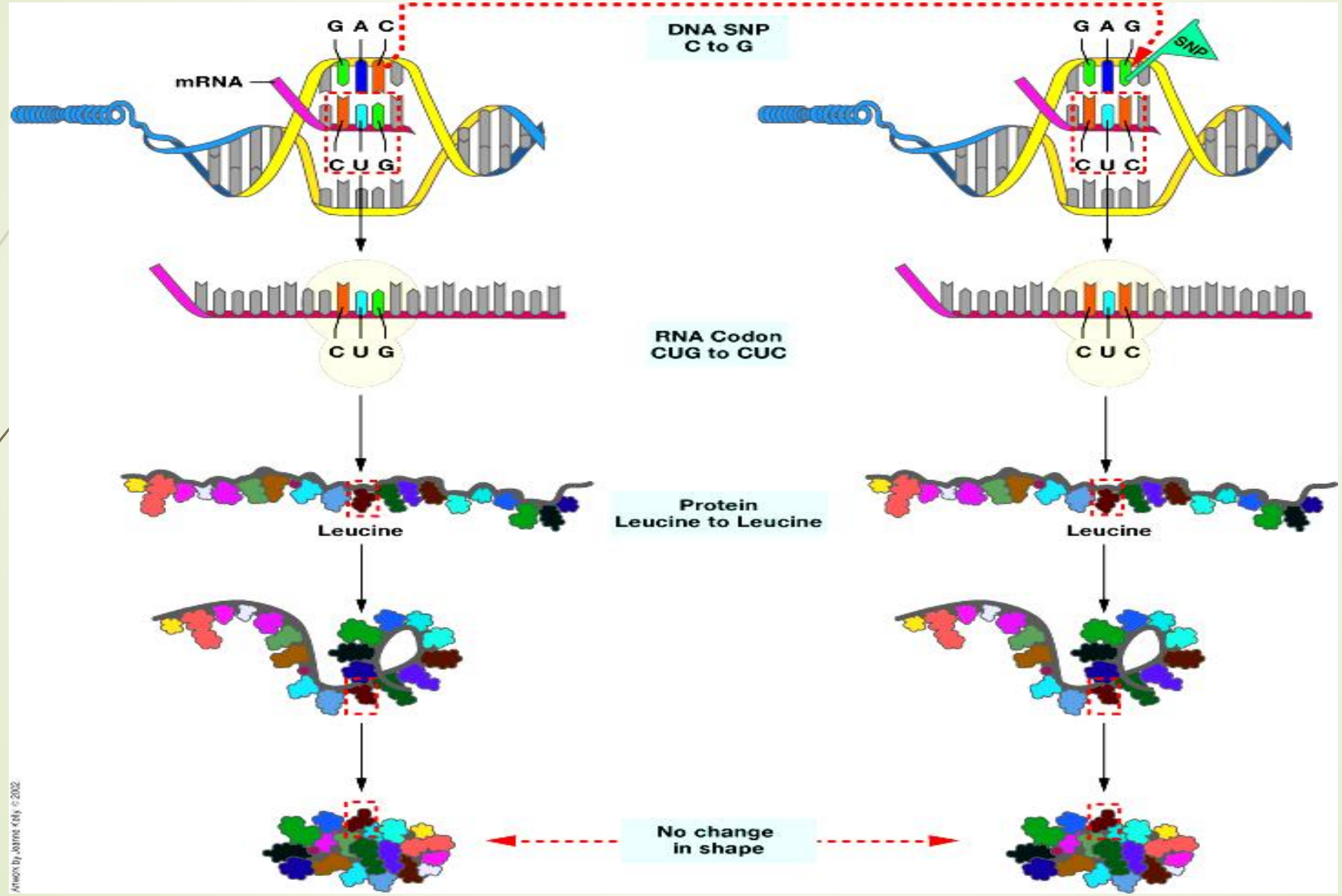
# Factors Influencing Mutation

- **Lifestyle Factors**
- Smoking: Smoking can increase the risk of mutation, particularly in lung cells.
- Diet: A diet high in processed foods and low in fruits and vegetables may increase the risk of mutation.
- Age: The risk of mutation increases with age due to the accumulation of genetic damage over time (Romero et al., 2022).
- **Random Errors**
- Replication errors: Errors during DNA replication can lead to mutations.
- Transcription errors: Errors during transcription can lead to mutations.
- **Epigenetic Factors**: Changes in epigenetic marks, such as DNA methylation or histone modification, can influence mutation rates.
- **Genomic Instability**: Conditions that lead to genomic instability, such as cancer or certain genetic disorders, can increase the risk of mutation.

# SINGLE NUCLEOTIDE POLYMORPHIMS (SNPs)

- Single Nucleotide Polymorphisms (SNPs) are the most common type of genetic variation found in the human genome. Each SNP involves a change of a single nucleotide — A, T, C, or G — at a specific position in the genome (Brooks et al., 2016).
- It occurs approximately every 100–300 base pairs and is present in both coding and non-coding regions of the genome.
- Most SNPs are biologically neutral, but some influence disease susceptibility (e.g., diabetes, Alzheimer's) and response to drugs (e.g., warfarin, clopidogrel).
- SNPs are inherited and serve as genetic markers in population studies and disease association studies.
- Example: Substitution of C with T in a gene regulatory region may alter gene expression.

# Some SNPs may or may not alter protein structure



# TYPES OF SNPs

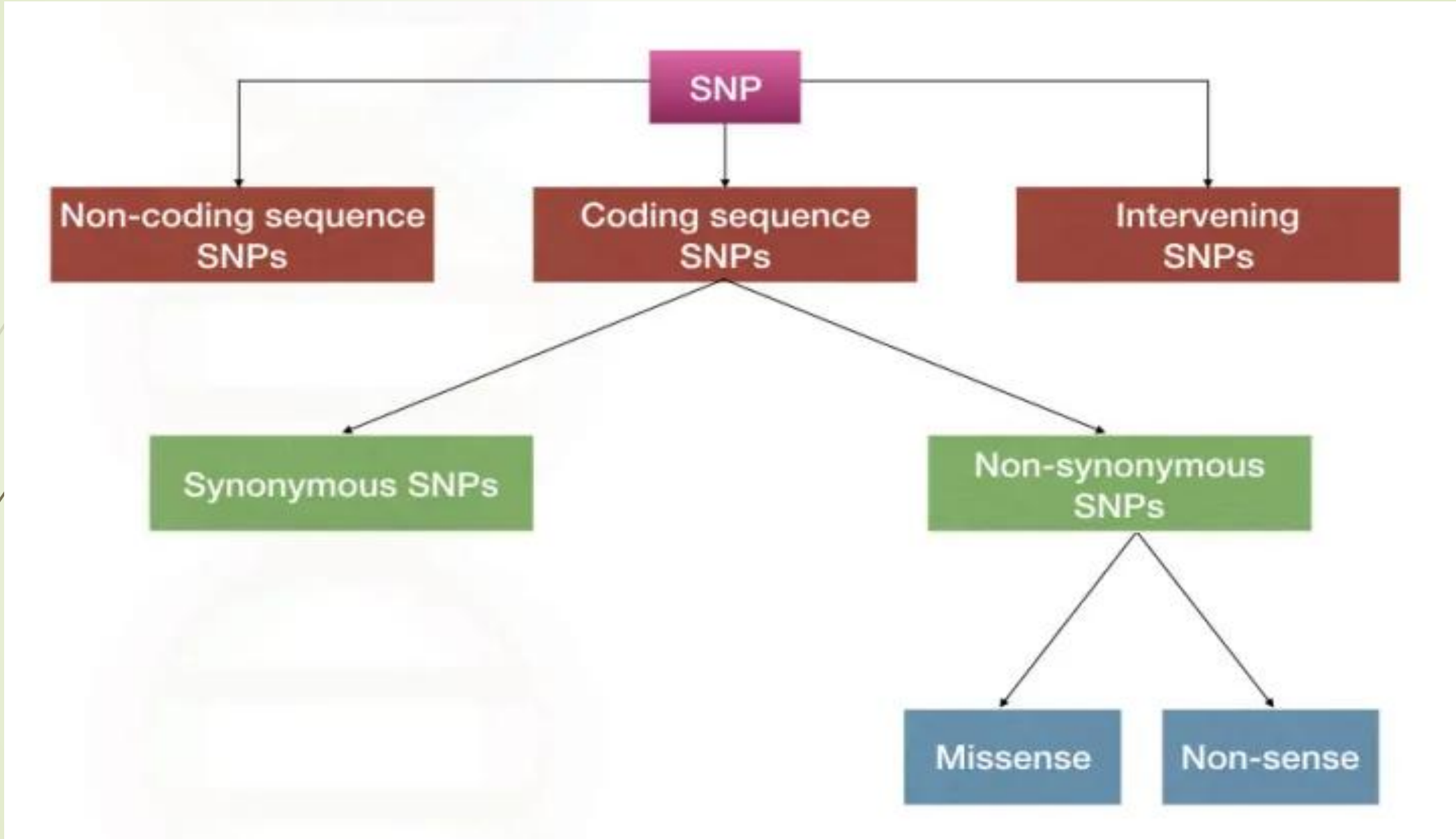
1. **Coding Region SNPs:**

- Synonymous (Silent): No change in the amino acid sequence. Example: GAA → GAG (both code for Glutamate).
- Non-synonymous (Missense): Alters a single amino acid (e.g., Glu → Val).
- Nonsense: Introduces a premature stop codon (e.g., UAU → UAA).

2. **Non-Coding Region SNPs:**

- Promoter or Enhancer SNPs: Affect transcription factor binding and gene expression.
- Splice Site SNPs: Disrupt mRNA splicing, leading to altered protein isoforms.
- UTR SNPs (5′ or 3′ Untranslated Regions): Influence mRNA stability, localization, and translation efficiency.

3. **Intergenic SNPs:** Located between genes. May affect regulatory elements, non-coding RNAs, or long-range chromatin structure


SNP
Non-coding sequence SNPs
Coding sequence SNPs
Intervening SNPs
Synonymous SNPs
Non-synonymous SNPs
Missense
Non-sense

# Techniques to detect known Polymorphisms

- **Hybridization Techniques**
  - Micro arrays
  - Real time PCR
- **Enzyme based Techniques**
  - Nucleotide extension
  - Cleavage
  - Ligation
  - Reaction product detection and display

# Techniques to detect unknown Polymorphisms

- Direct Sequencing
- Microarray
- Cleavage / Ligation
- Electrophoretic mobility assays

# Significance of SNPs

- In Disease Diagnosis
- In Finding Predisposition to Diseases
- In Drug Discovery & Development
- In Drug Responses
- Investigation of Migration Patterns

# REFERENCES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