

Al-Mustaqbal College University

Department Of Dentistry

Medical Biology



Mutation

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mutation

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graph LR; A[mutation] --> B[Gene mutation]; A --> C[Chromosome mutation]; B --> D[Changes in the structure of DNA]; C --> E[Changes of number or structure of whole chromosome]
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Gene mutation

Changes in the structure of DNA

Chromosome mutation

Changes of number or structure of whole chromosome

Gene mutation

Base substitution

- One base replaces another
- CCTG**A**GGAG → CCTG**T**GGAG
- Silent mutation

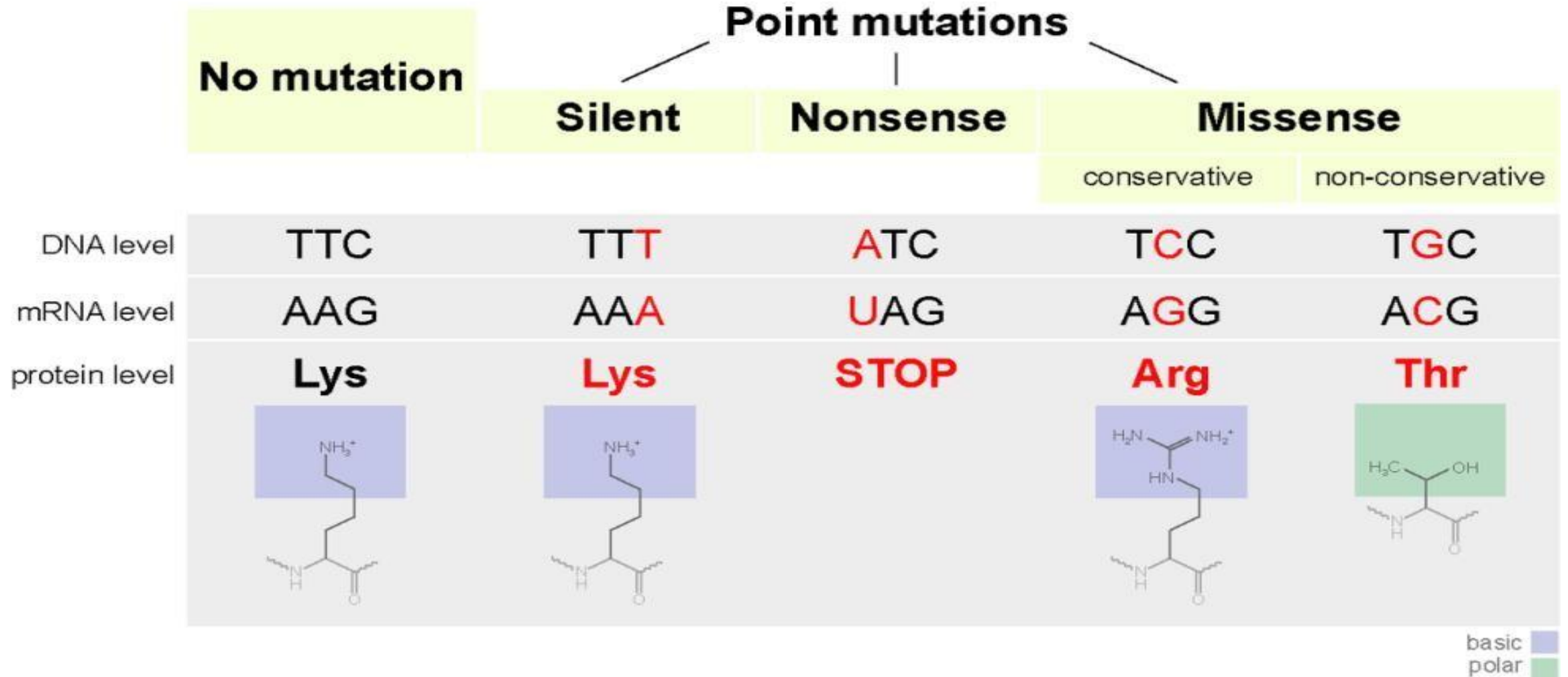
Base addition

- One or more extra bases are added
- CCAGAGGGAG → CCA**A**GAGGGAG
- Frame shift

Base deletion

- One or more bases are deleted
- CCTGAGGGAG → CCTGAGAG
- Frame shift

Point Mutations

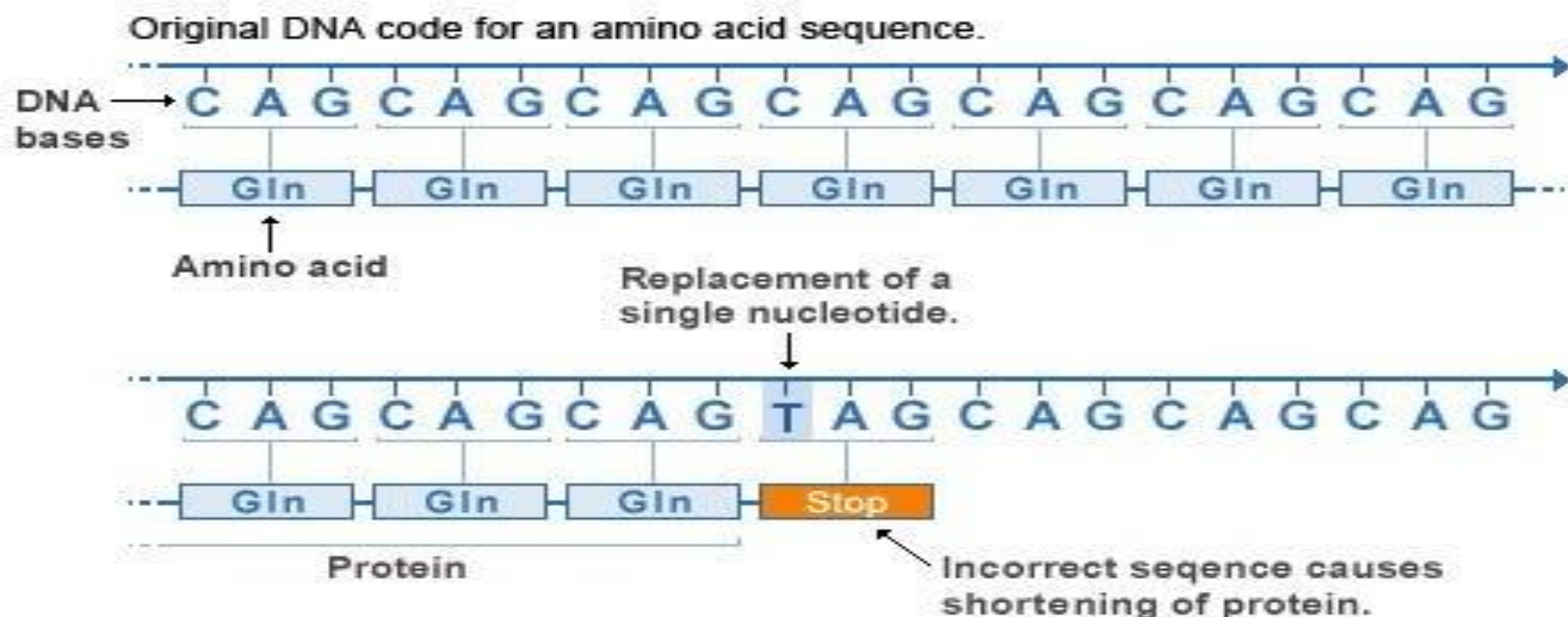


- Point mutations are changes in one base pair of a cell's DNA sequence.
- For example, if an A in the DNA code is changed to a C, that is a point mutation.
- Point mutations in the coding region of a gene can have different effects depending on the resulting changes to the codons in the messenger RNA.
- There are a few major kinds of point mutations: missense mutations, nonsense mutations, silent mutations and read through mutations.
- Here, we're going to focus on nonsense mutations, which are mutations that introduce a premature stop codon into the coding sequence of a gene.

Nonsense Mutation Definition

- When you think about a mutant, you might think about sci-fi movies where mutated creatures become powerful and evil and then attempt to destroy the world.
- But what are mutations, really?
- Mutations are changes to a cell's DNA sequence, and there are several different types.
- A **nonsense mutation** is a point mutation that introduces a premature stop codon into the part of the gene that encodes a protein.

Nonsense mutation



Gene vs. Chromosomal Mutations

Gene Mutations

- Mistakes that affect individual genes on a chromosome.
- One base substitutes for another on a DNA strand and leads to the wrong protein being made; this affects one or more functions within the organism.

Chromosomal Mutations

- Mistakes that affect the whole chromosome.
- There are four types of chromosomal mutations: duplication, deletion, inversion, and translocation.
- ALL MUTATIONS ARE CAUSED BY MUTAGENS.

- A **chromosome abnormality, disorder, anomaly, aberration,**
- or **mutation** is a missing, extra, or irregular portion of [chromosomal](#) DNA.

Change of chromosome structure

Duplication

Genes sequences are **repeated** several to many hundreds or thousands of times.

Deletion

Loss of some segment of a chromosome

Inversion

A linear segment of DNA within the chromosome becomes oriented in the **reverse direction**, with no molecular loss.

Translocation

Exchange of segments DNA between **non-homologous chromosomes**

Change of chromosome number

Changes number in autosome chromosomes

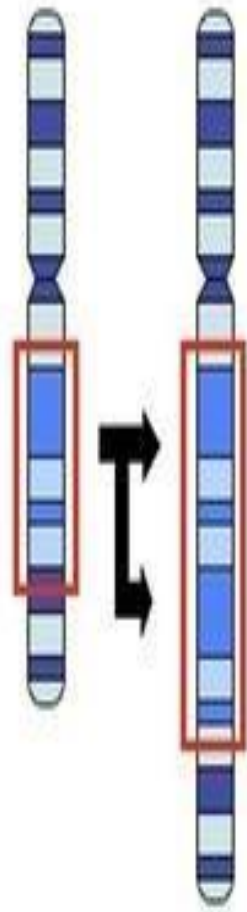
Example: Down's syndrome

Changes number in sex chromosomes

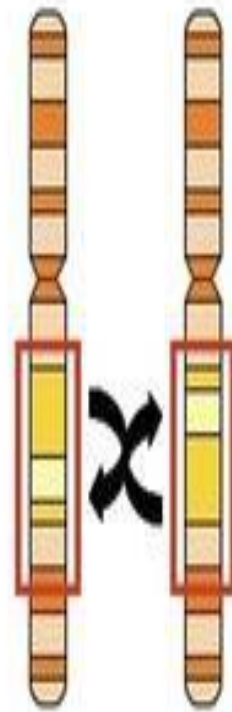
Examples: Turner syndrome, Klinefelter syndrome, XYY condition.

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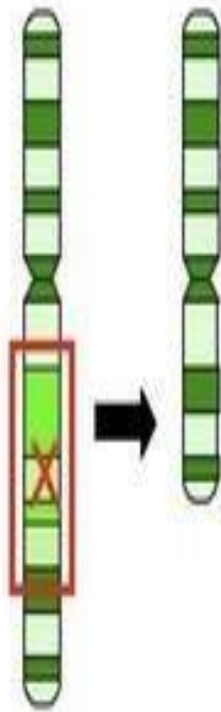
Duplication



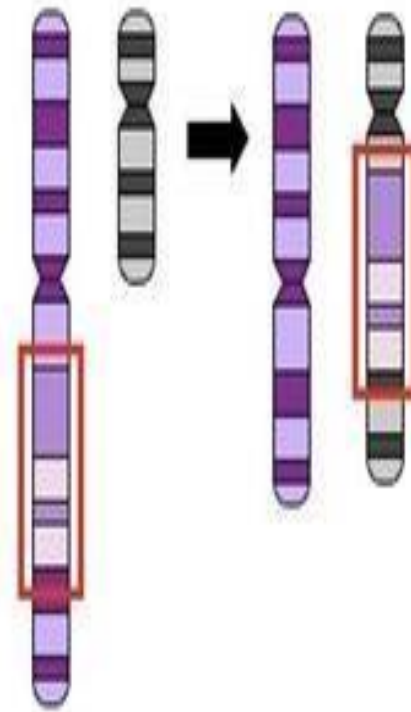
Inversion



Deletion



Insertion



Translocation

