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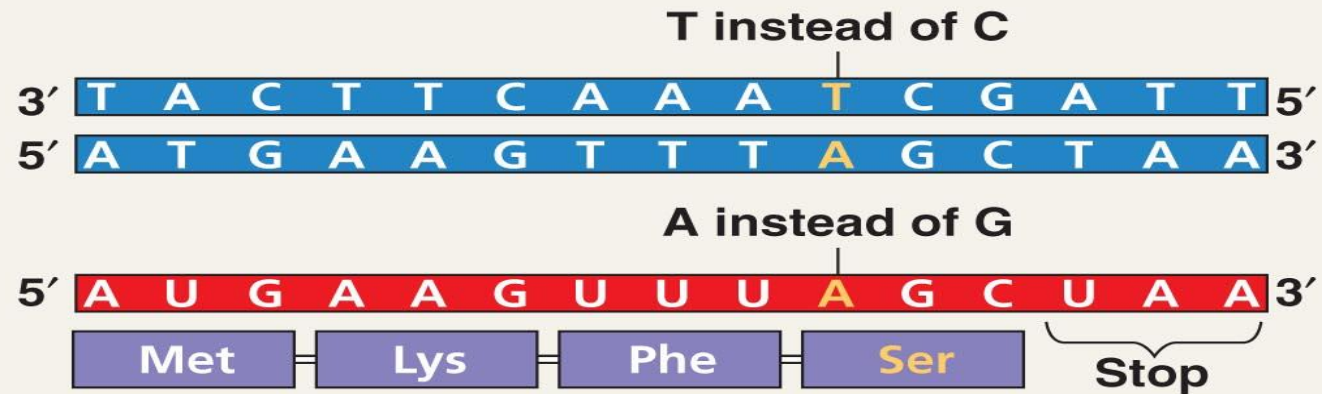
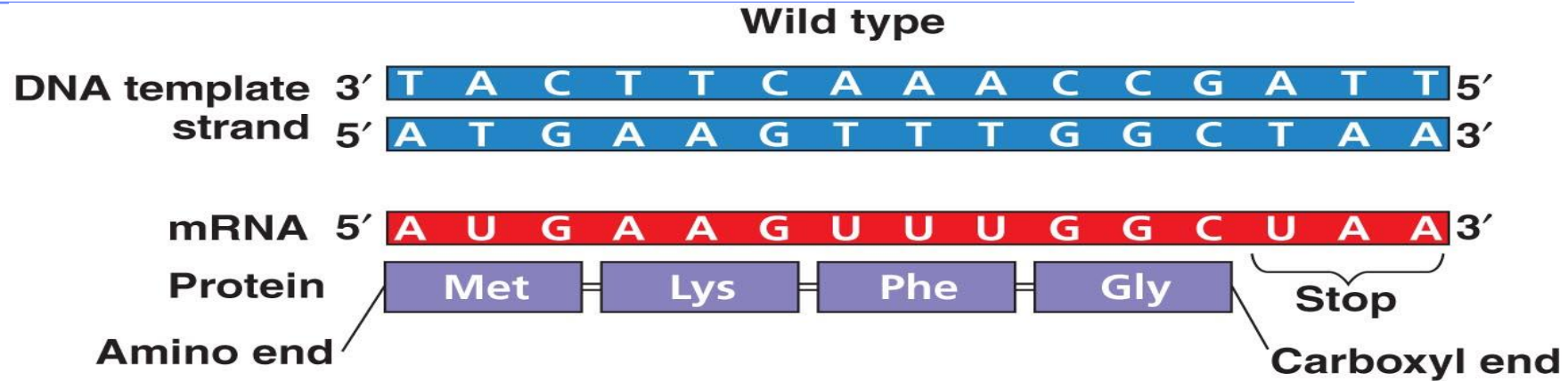
Chemistry department

**Transcription of DNA's Genetic
Information, Genetic Mutations and
Factors Causing Genetic Mutations**

Mutations = changes in the genetic material of a cell

- Large scale mutations: chromosomal; always cause disorders or death
 - ◆ nondisjunction, translocation, inversions, duplications, large deletions
- Point mutations: alter 1 base pair of a gene
 1. Base-pair substitutions – replace 1 with another
 - Missense: different amino acid
 - Nonsense: stop codon, not amino acid
 2. Frameshift – mRNA read incorrectly; nonfunctional proteins
 - Caused by insertions or deletions

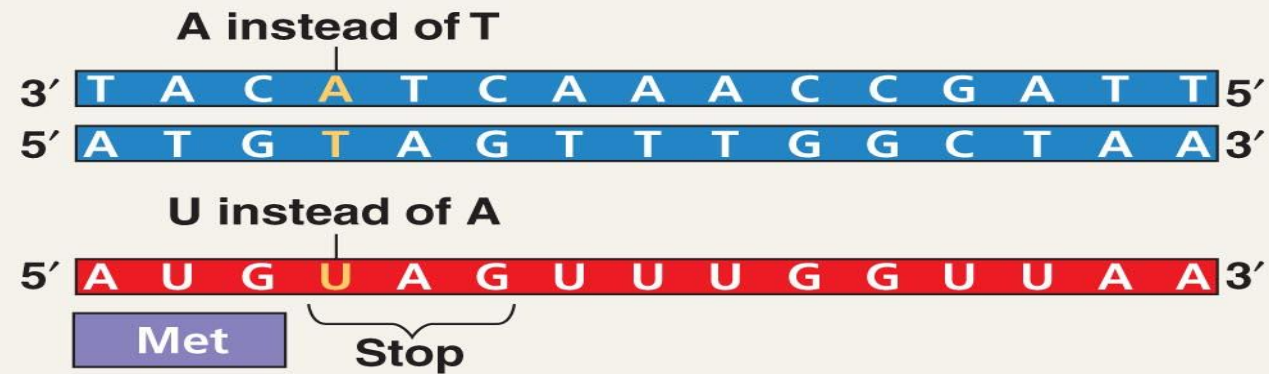
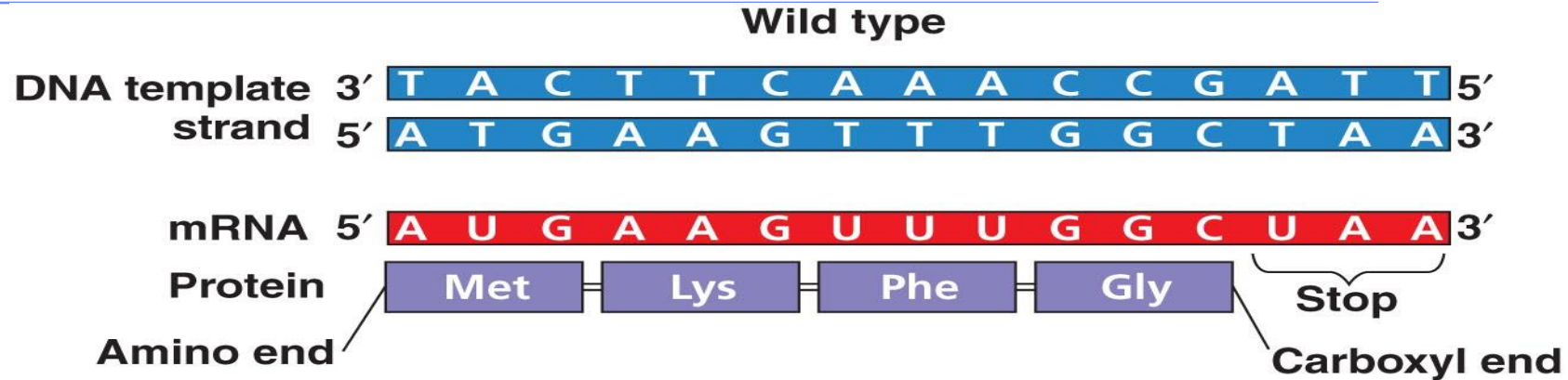
Substitution = Missense



Missense

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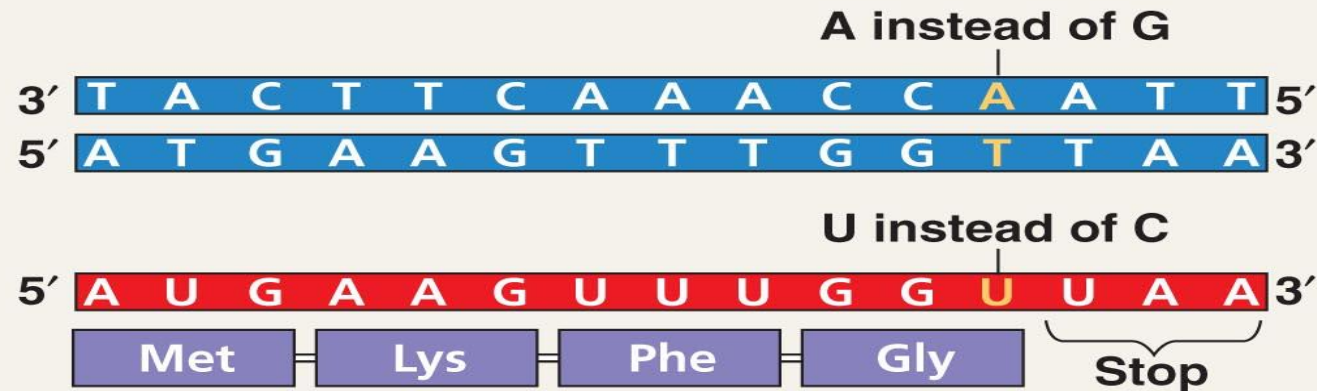
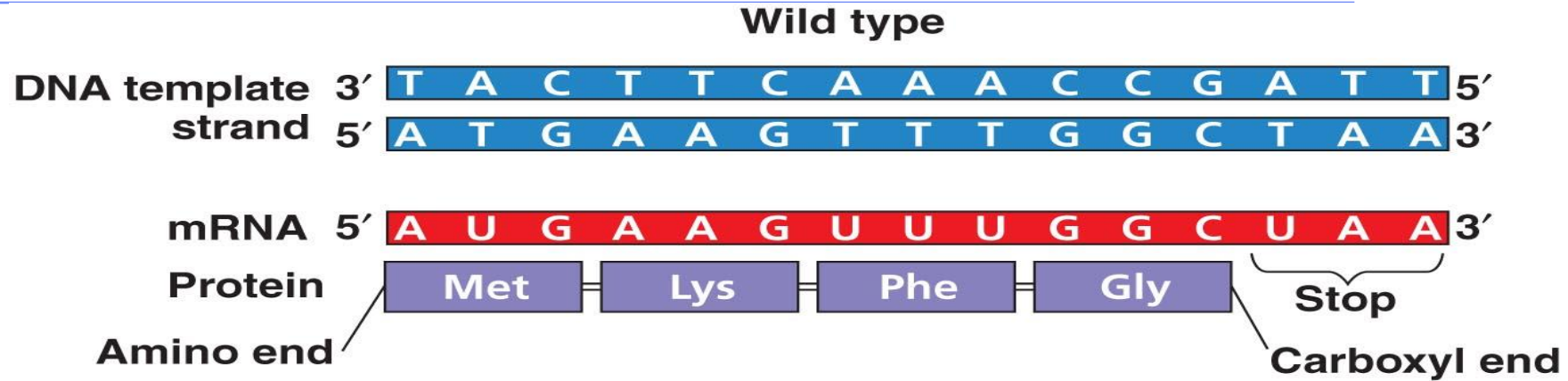
Substitution = Nonsense



Nonsense

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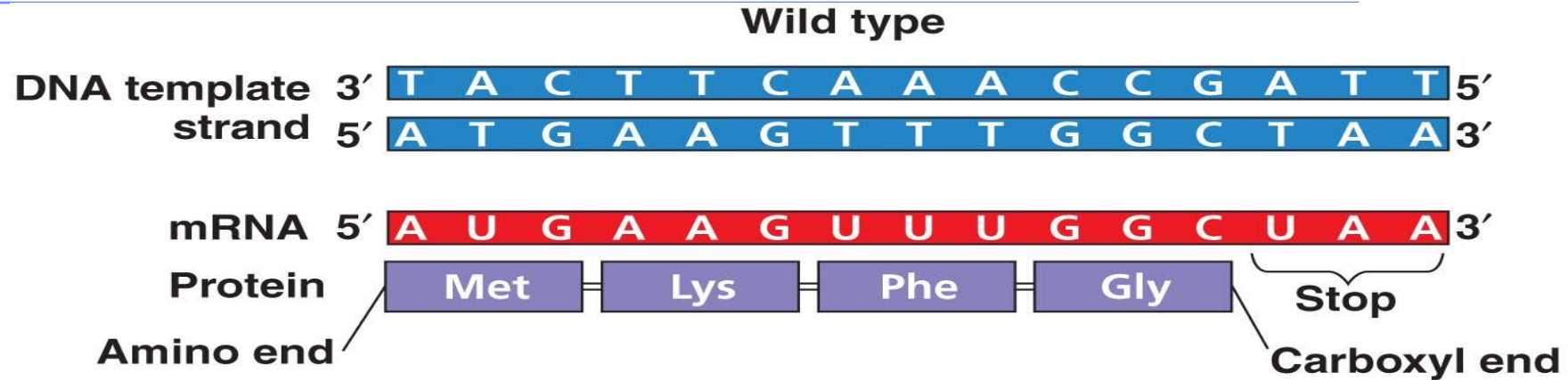
Substitution = Silent (no effect)



Silent (no effect on amino acid sequence)

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Insertion = Frameshift Mutation

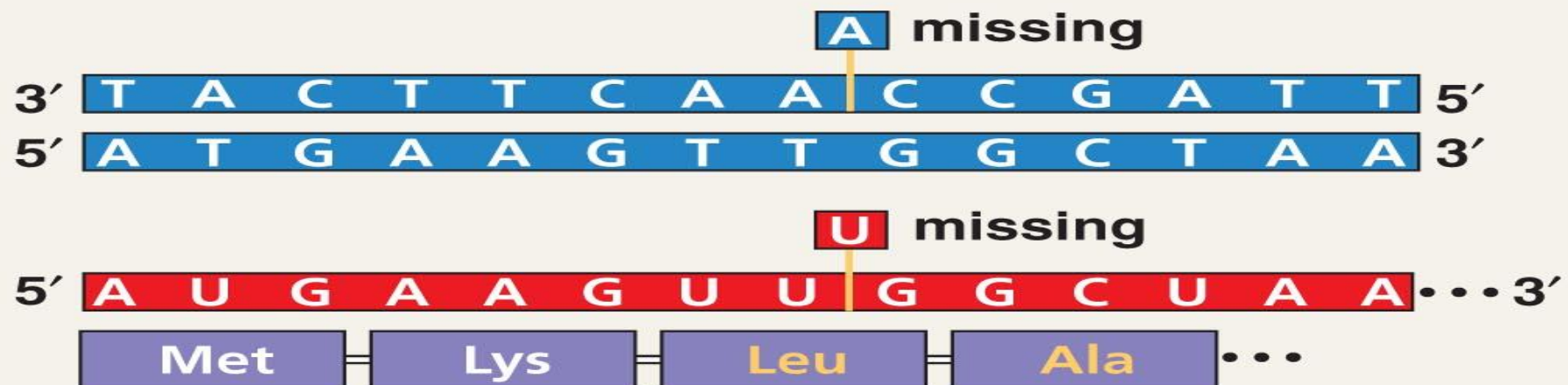
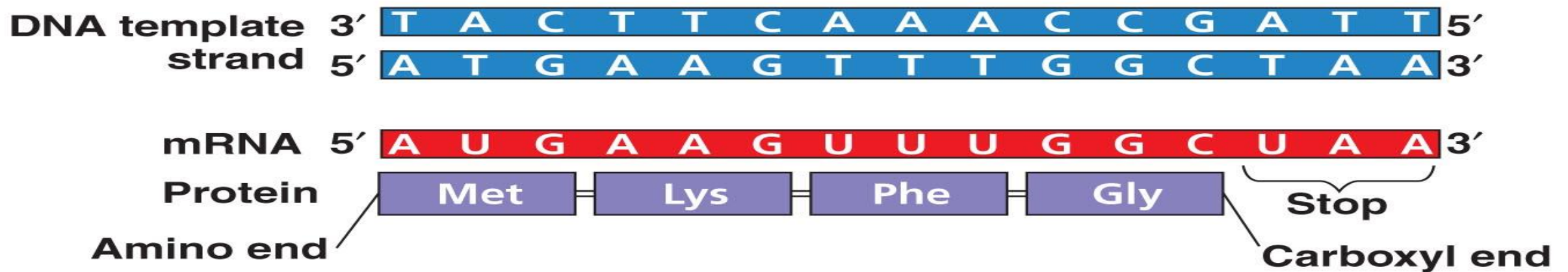


Frameshift causing immediate nonsense (1 base-pair insertion)

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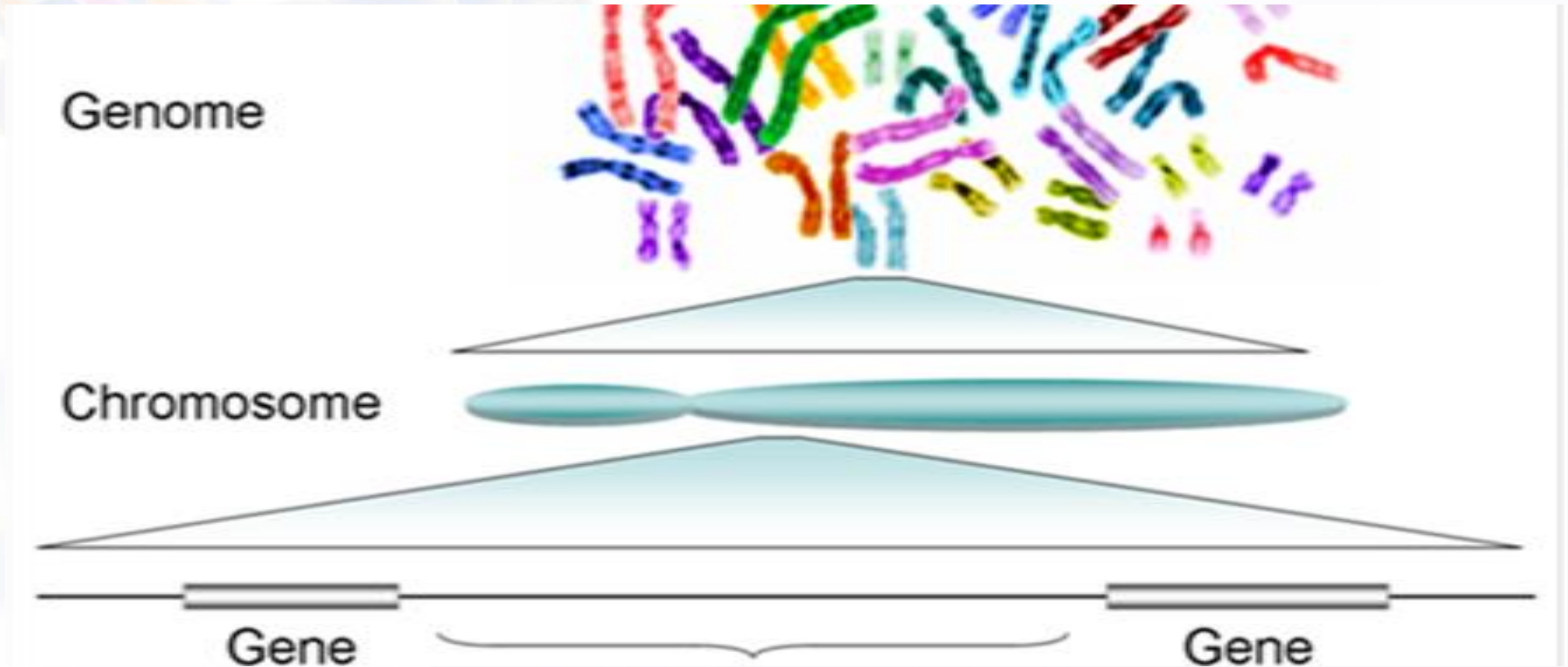
Deletion = Extensive missense, premature termination

Wild type



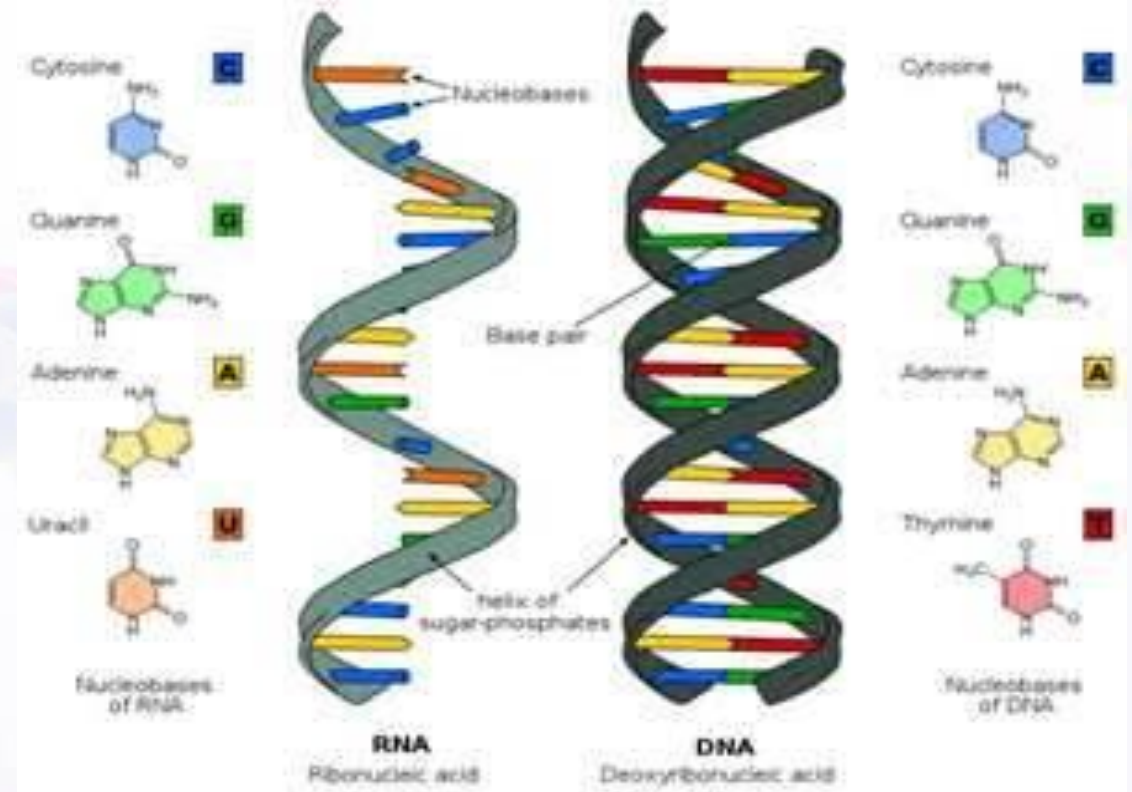
Frameshift causing extensive missense (1 base-pair deletion)

Human Genome



General Structure of Nucleic Acid

DNA and **RNA** are long chain polymers of small chemical compound called nucleotides.



Nucleotides

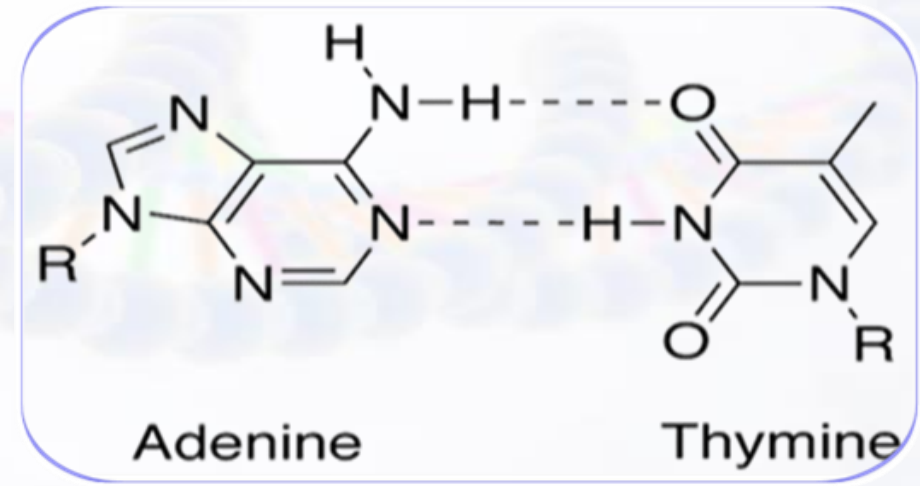
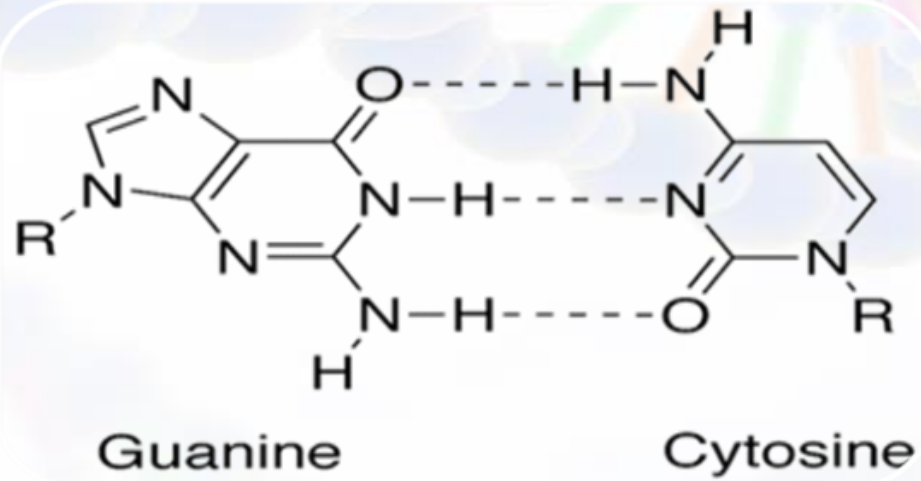
Nucleotides; ring shaped structures composed of:

- Nitrogenous base; these bases are classified based on their chemical structures into two groups:
 - Purine; double ringed structure (Adenine and Guanine).
 - Pyrimidine; single ring structures (cytosine and thymine).
- Sugar
- Phosphate group

Nucleotides

- **DNA:** Four different types of nucleotides differ in nitrogenous base:
 - A is for adenine;
 - G is for guanine;
 - C is for cytosine and
 - T is for thymine.
- **RNA:** thymine base replaced by uracil base.

Nucleotides



The DNA

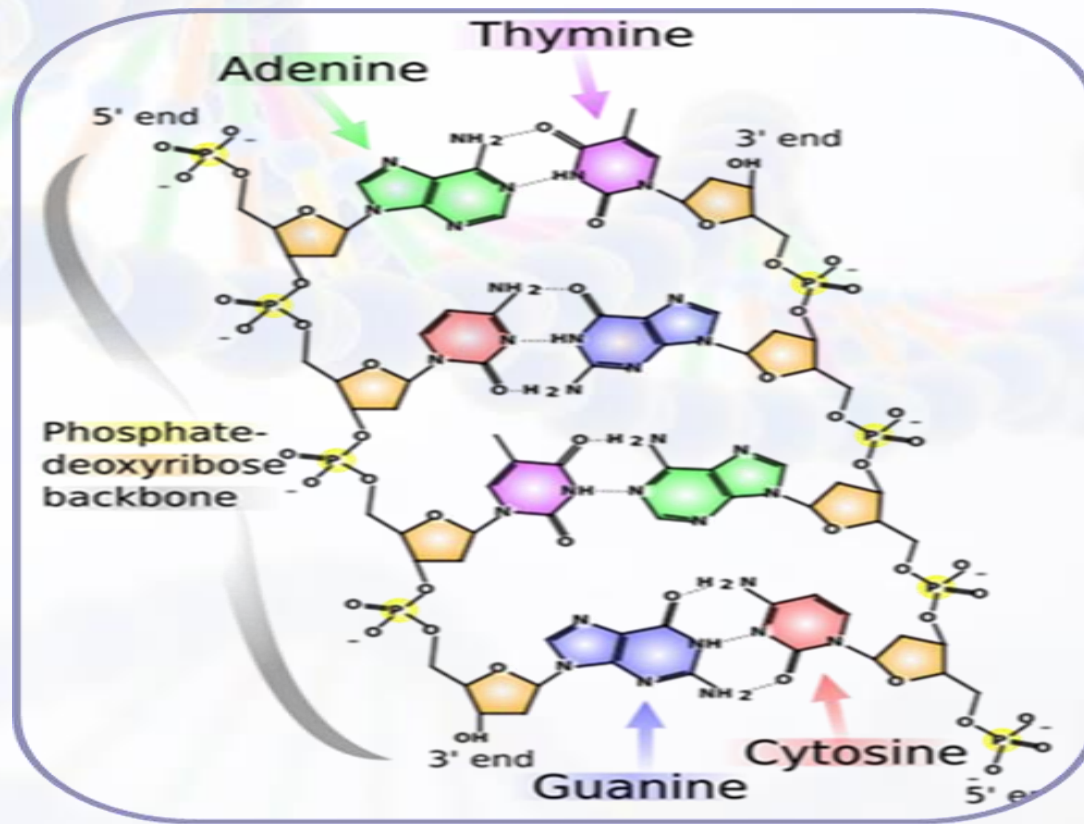
- **Deoxyribonucleic Acid (DNA)**; the genetic material of all cellular organisms and most viruses.
- **DNA**; the gigantic molecule which is used to encode genetic information for all life on Earth.
- A human cell contains about 2 meters of **DNA**. **DNA** in the body could stretch to the sun and back almost 100 times. So it is tightly packed.
- **DNA** responsible for preserving, copying and transmitting information within cells and from generation to generation.



DNA Double Helix

- Linked as a twisted ladder.
- The curving sides of the ladder represent the **sugar-phosphate** backbone of the two DNA strands; the rungs are the **base pairs**.
- Possess **antiparallel** polarity.
- Stabilized by **hydrogen bonds** between the bases.

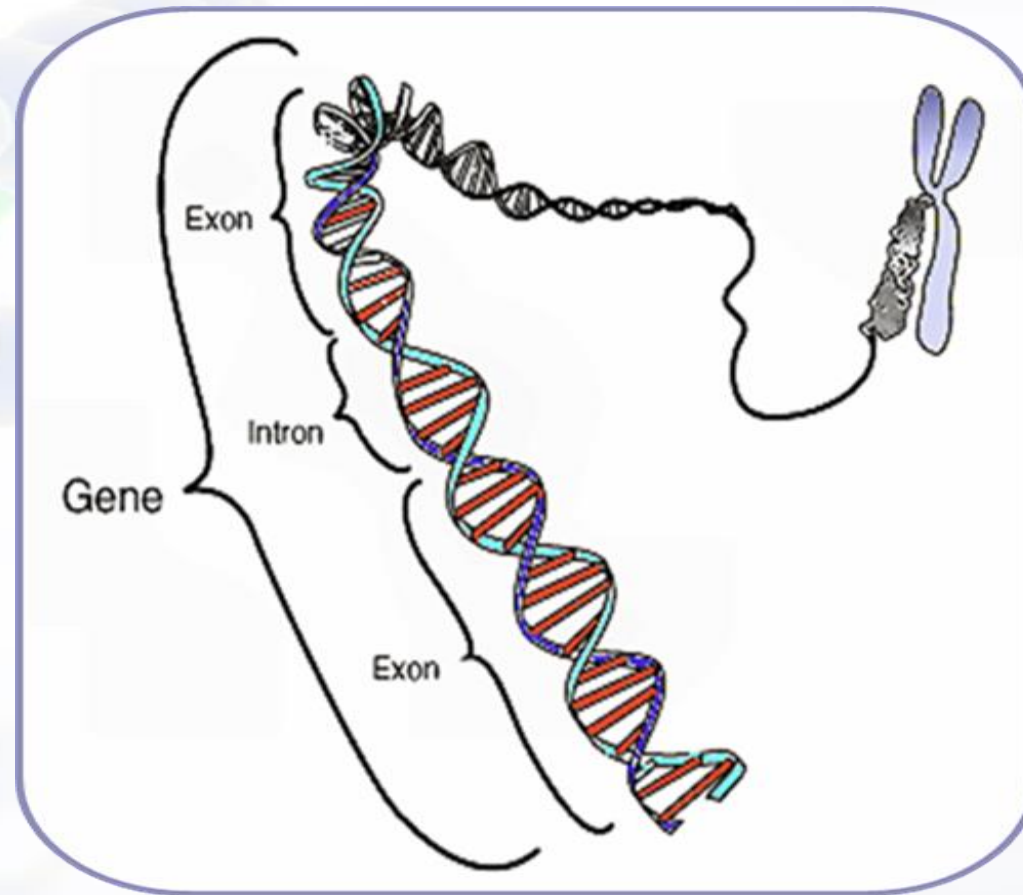
DNA Double Helix



The Gene

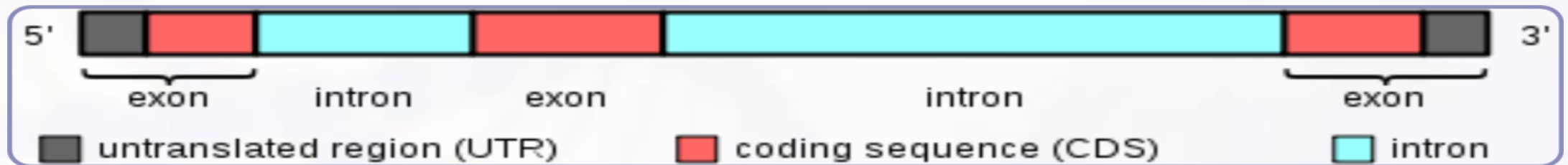
- **The gene**; it is a segment within a very long strand of **DNA**.
- **Genes** are the basic units of hereditary.
- **Genes** located on chromosome on its place or **locus**.
- **Allele**; a variant of the DNA sequence at a given locus. Each allele inherited from a different parent.

The Gene



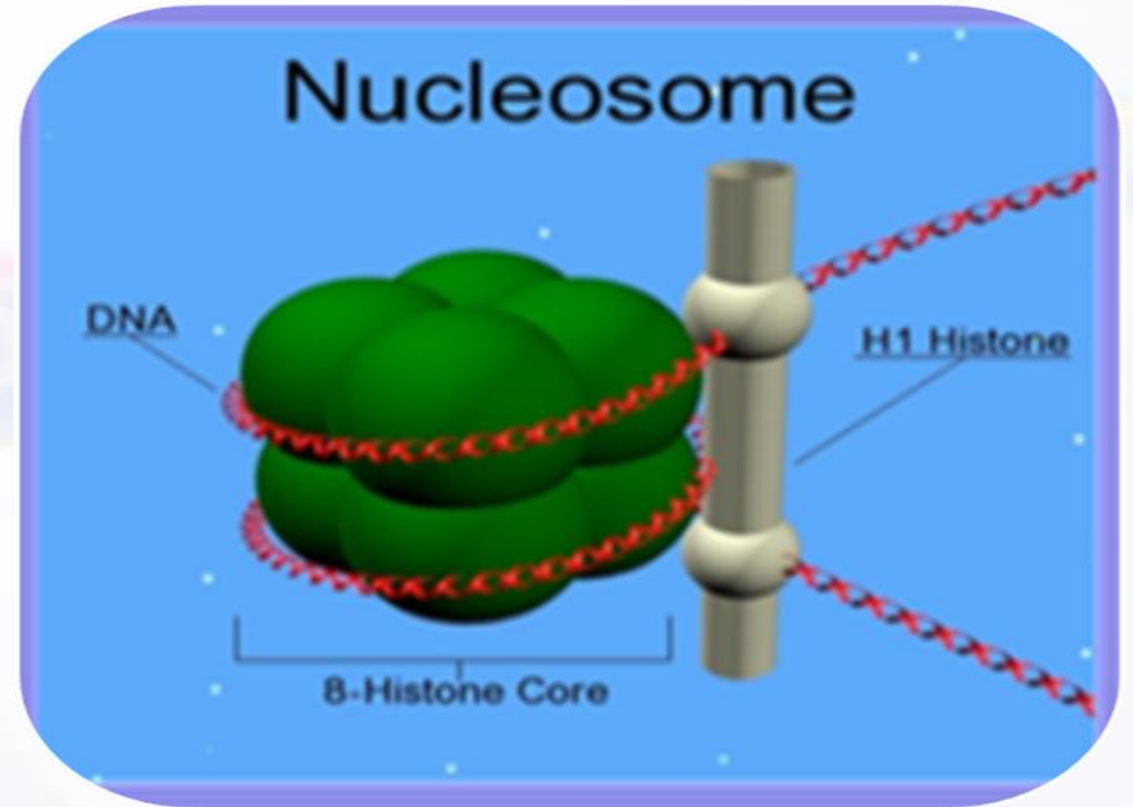
Gene Structure

- Most of the genes consist of; short coding sequences or exons are interrupted by a longer intervening noncoding sequence or introns; although a few genes in the human genome have no introns.

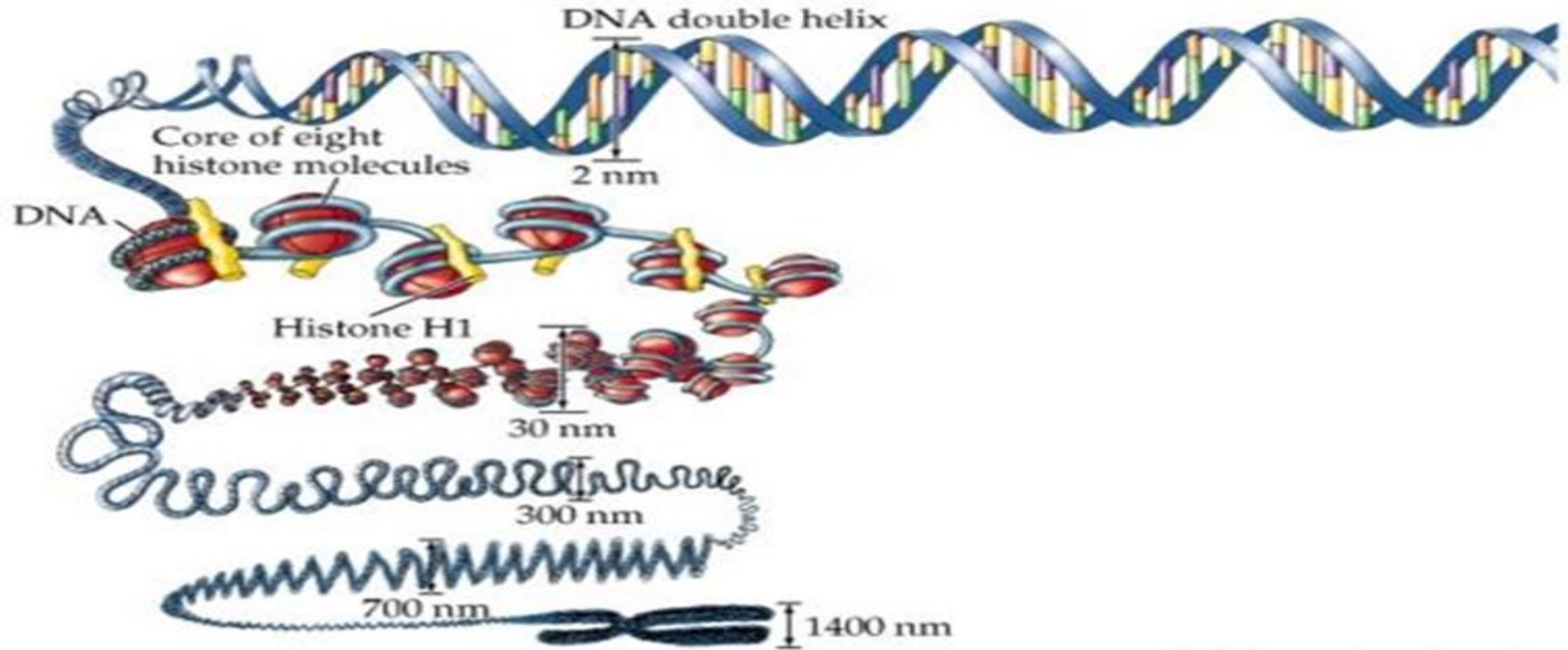


DNA Organization

DNA molecules complexed with other proteins, especially basic proteins called histones to form a substance known as chromatin.



DNA Organization



Cause and effect of mutation

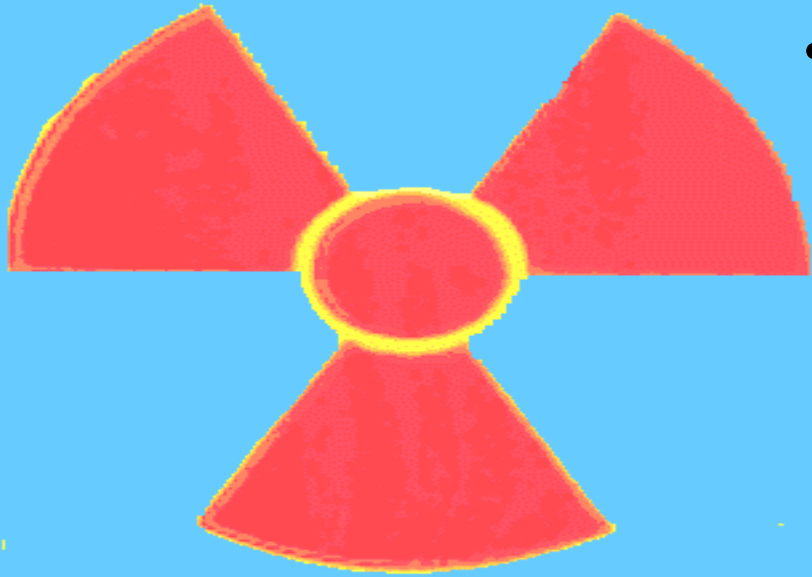
What causes mutation

- Spontaneous
- Increases caused by environmental factors
- UV light
- X-rays
- Benzene, formaldehyde, carbon tetrachloride

Gametic and somatic mutations

- Gametic - testis of males, ovaries of females, inherited
- Somatic - in normal body cells occurring beyond zygote formation, not inherited but may effect the person during their lifetime. Chimaeras

Mutagens and their effects



- Ionising radiation - Nuc radiation, xrays, gamma rays (e.g. medical treatment) associated with development of cancers (e.g. leukaemia, thyroid cancer and skin cancer)

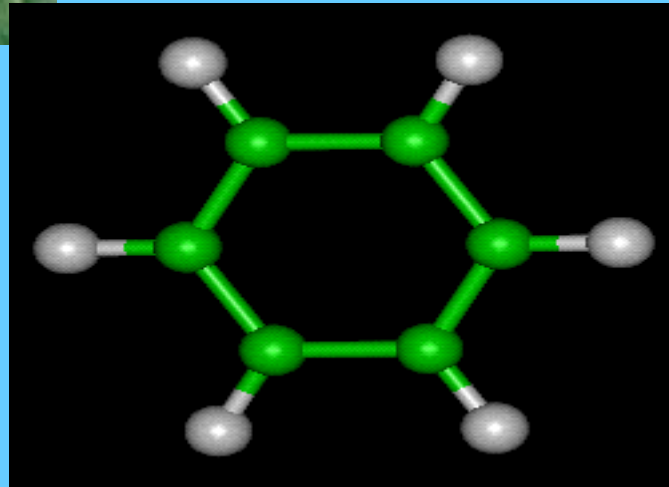
Mutagens and their effects



Plate 21 *Aspergillus*, the Green Mold, growing on malt agar media.

- Viruses and microorganisms - integrate into human chromosome, upset genes and can trigger cancer

Mutagens and their effects



- Environmental poisons
 - Organic solvents such as formaldehyde, tobacco, coal tars, benzene, asbestos, some dyes

Mutagens and their effects



- Alcohol and diet - High alcohol intake increase the risk of some cancers. Diet high in fat and those containing burned or highly preserved meat



The effect of mutations

- Not all are harmful
- Survival advantage
- Most common among bacteria and viruses but also seen in insects
- If no selective pressure may remain in population

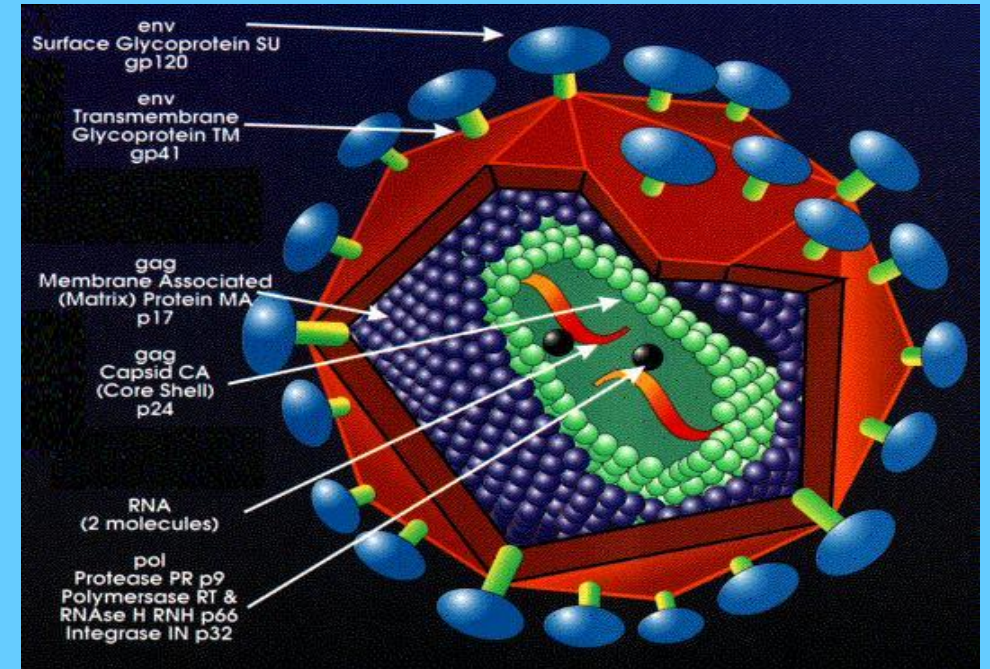
Harmful mutations

- Cystic fibrosis and sickle cell anaemia
- Disfunctional proteins
- Albinism - caused by mutation in gene of enzyme pathway of melanin



Beneficial mutations

- Bacteria - antibiotic resistance through mutation, transfer between bacterial species
- Superbugs such as MRSA have arisen this way
- RNA viruses - such as HIV - mutates it's protein coat so that the host human is unable to make antibodies quick enough against it



Neutral mutations

- Neither harmful or beneficial to the organism but may be important in an evolutionary sense
- Silent mutations
- Virtually impossible to detect because no observable effect

Thank you