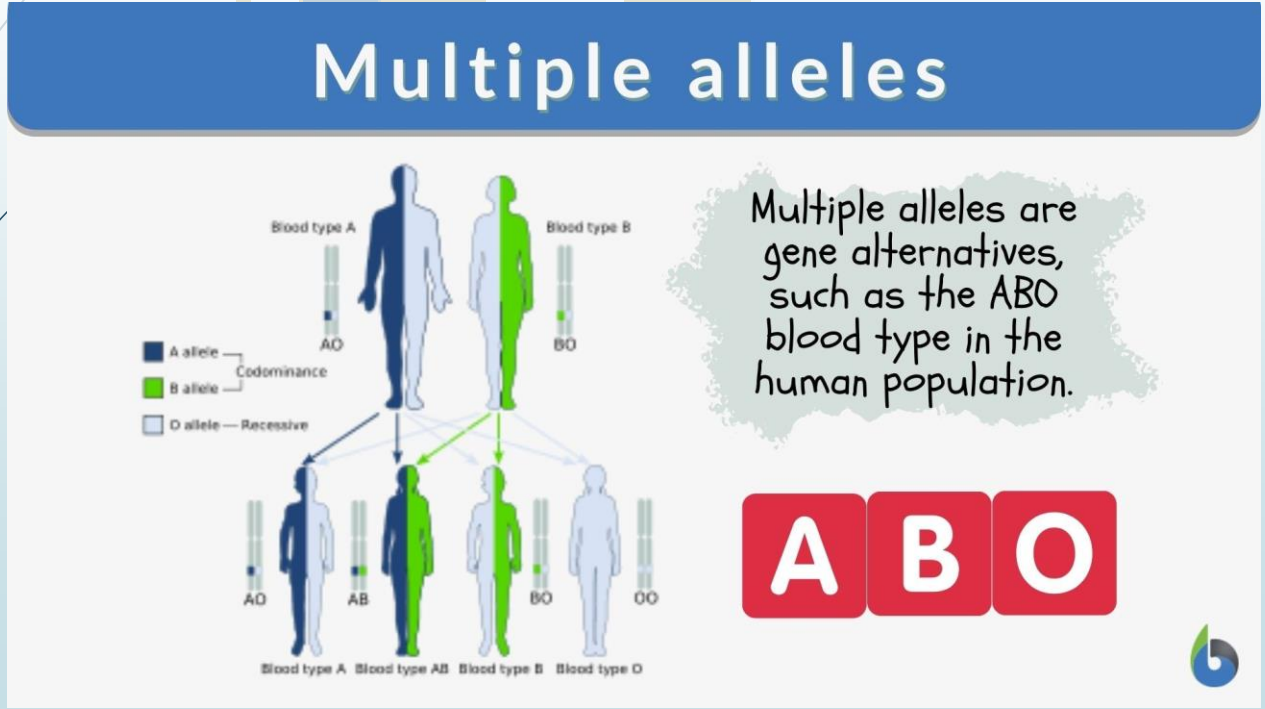
Document 2

Mutation and Multiple Alleles

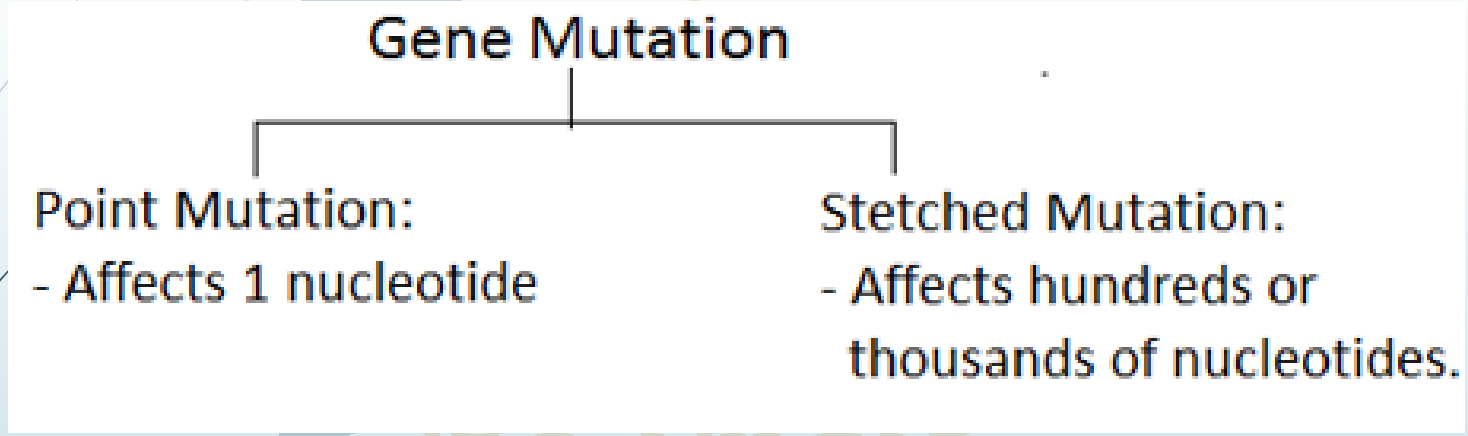


- Mutation: is a change in the nucleotide sequence of DNA.
- **Mutations may occur in genes or chromosomes:**

- Gene mutation: is a change in the nucleotide sequence of the DNA within a gene.
- Chromosome mutation: affects the number or structure of chromosomes.(To be discussed in chapter 5)

- DNA has coding (30,000 genes per diploid cell) and non-coding sequence of nucleotides (95% of the DNA).
- **Only** mutations in the coding region of the DNA may alter the function of the protein.

I. Types of Gene Mutation



- **Types of Point Mutations:**

Doc.b, p.60, shows different types of point mutations and their consequences.

type of mutation	non-transcribed DNA strand of a normal gene and corresponding amino acids	non-transcribed DNA strand of the mutant gene and corresponding amino acids	effect of the mutation
substitution	....CCA-GAG-ACT.... Pro - Glu - Thr	....CCA-GTG-ACT.... Pro - Val - Thr	missense altered polypeptide
	....CCA-GAG-ACT.... Pro - Glu - Thr	....CCA-GAA-ACT.... Pro - Glu - Thr	silent no detectable change
	....CCA-GAG-ACT.... Pro - Glu - Thr	....CCA-TAG-ACT.... Pro - Stop -	nonsense incomplete polypeptide
deletion	....TAC-ACC-ACG-A... Tyr - Thr - Thr	....TAC-CCA-CGA... Tyr - Pro - Arg	frame-shift altered polypeptide
insertion	....TAC-ACC-ACG-A... Tyr - Thr - Thr	....TAC-GAC-CAC-GA... Tyr - Asp - His	frame-shift altered polypeptide

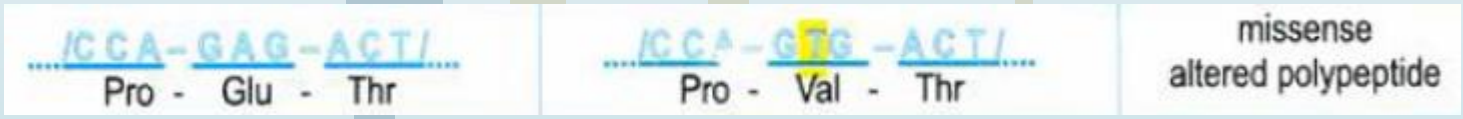
Doc. b *Main types of point mutations and their consequences. The non-transcribed DNA strand (coding strand) gives a quick and direct reading of the messenger RNA by just replacing thymine (T) with Uracil (U).*

1- Substitution: One nucleotide is replaced by another.

➢ Consequence (effect):

1.1- Missense mutation: GAG (Glu) → GTG (Val)

- The change in 1 nucleotide leads to a **different codon** which codes for a different amino acid.



⇨the polypeptide (protein) is altered.

⇨the phenotype will be affected.

1.2- Silent Mutation: GAG (Glu) → GAA (Glu)

- The change in 1 nucleotide gives a codon that codes for the same amino acid .

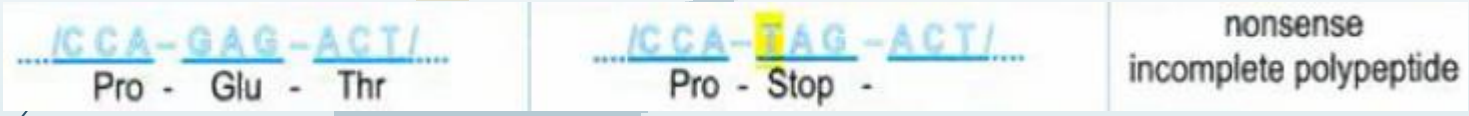
....CCA-GAG-ACT.... Pro - Glu - Thr	....CCA-GAA-ACT.... Pro - Glu - Thr	silent no detectable change

⇨no change in protein.

⇨the phenotype will not be affected.

1.3- Non-sense Mutation: GAG (Glu) → TAG (stop)

- The change in 1 nucleotide causes the formation of a stop codon.



⇨ formation of incomplete (truncated) non-functional protein.

⇨ phenotype will be affected.

2- Deletion: One nucleotide is removed.

➢ Consequence:

- Frame shift (backward): production of altered protein ⇨ affecting the phenotype.

3- Insertion: One nucleotide is added.

➢ Consequence:

- Frame shift (forward): production of altered protein ⇨ affecting the phenotype.

insertion	....TAC–ACC–ACG - A/... Tyr - Thr - Thr	....TAC–GAC–CAC - GA/... Tyr - Asp - His	frame-shift altered polypeptide

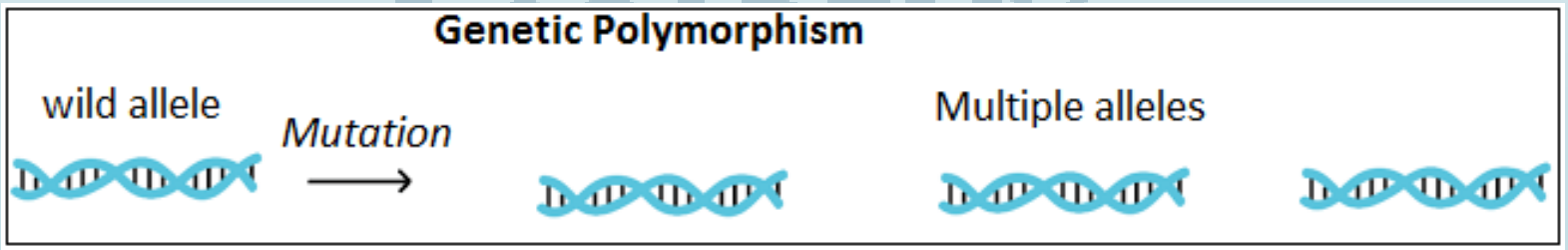
II. Genes and Multiple Alleles:

- Genetic polymorphism: is the presence of many alleles for the same gene, this gene is called polymorphic gene).

  example: blood type, eye color..

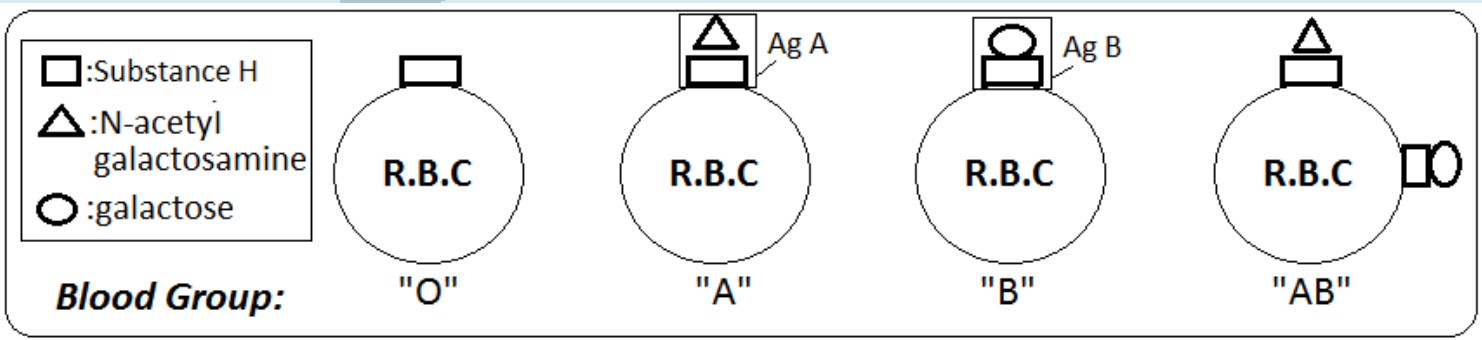
- **Cause of genetic polymorphism:** mutation of the wild allele (normal allele).

  -wild-type allele: is the allele that codes for the most common phenotype in a population.



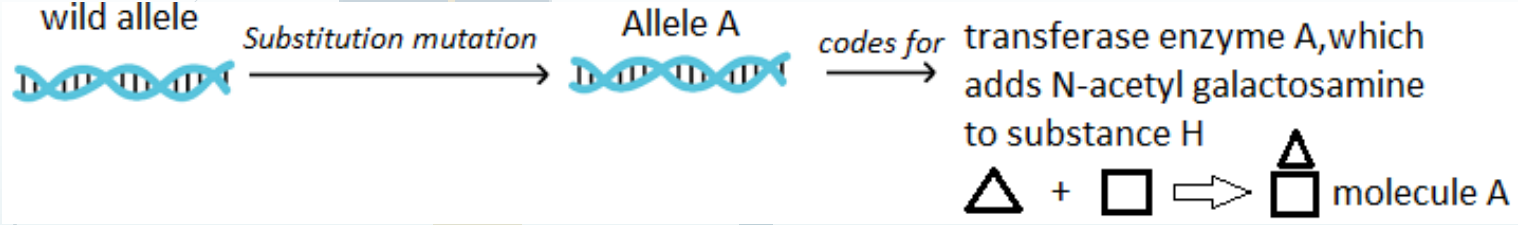
- Example of multiple alleles in humans is : ABO blood group system

- ABO system has three different alleles: A, B and O.
- It is characterized by the presence of O, A or B molecules on the surface of red blood cells.
- O, A and B molecules all have the same basic component called substance H, but they differ by the presence or absence of a sugar molecule.
- Document 1 shows a schematic diagram for different blood groups having different molecules O, A and B.

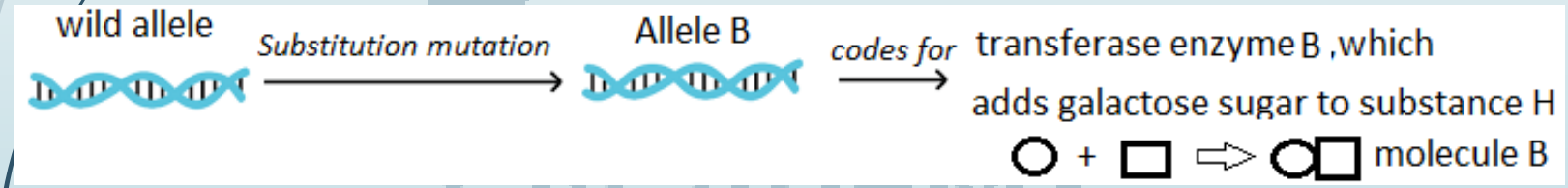


Document 1

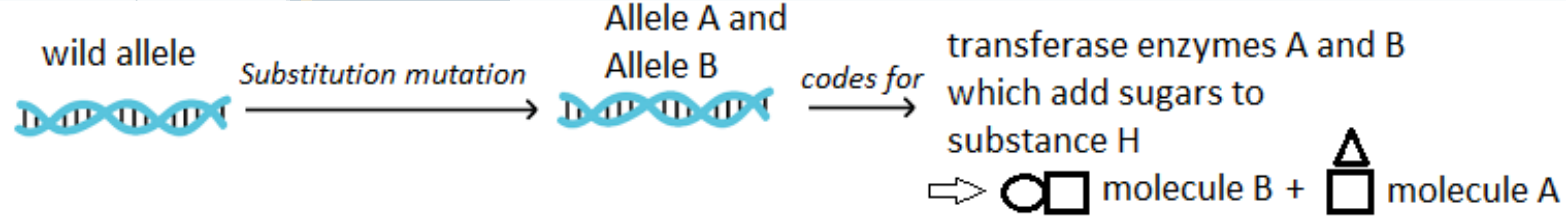
→Blood group A:



→ Blood B



→ Blood group AB:



→Blood group O: