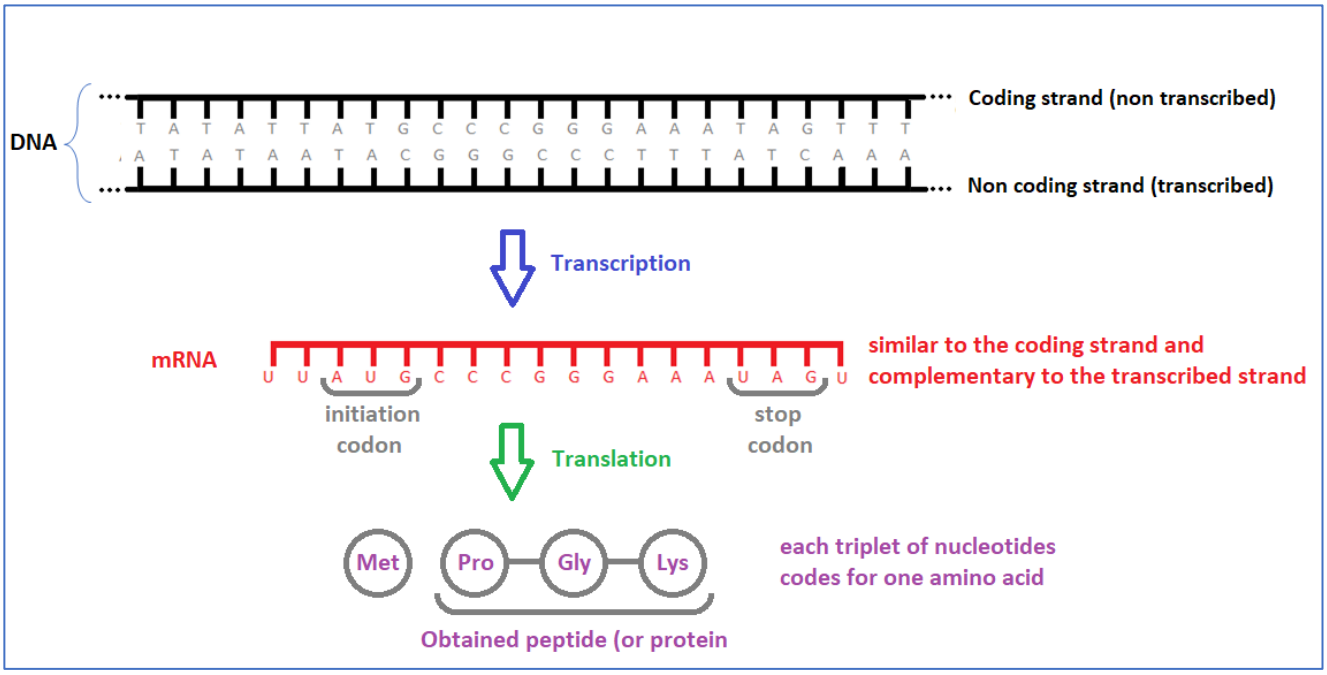
Ch.3 Act.2 Mutations and multiple alleles

Review:

- Genetic mutation: Change in the nucleotide sequence of the DNA.
- Only mutations in the germ cells can be transmitted to the offspring.
- Transcription of the DNA and translation of the obtained mRNA:



Types of genetic mutations:

Mutations, which affect a single nucleotide, are called point mutations. More stretched mutations, which affect hundreds or thousands of nucleotides can also occur.

The main types of mutations in the nucleotide sequence of the DNA are:

- Substitution: replacement of one or more nucleotide
- Deletion: elimination of one or more nucleotide
- Insertion: addition of one or more nucleotide

To identify a mutation, we indicate the change, its position and the name of the nucleotide.

Examples:

The nucleotide **G** number 35 is replaced by **A**
The second nucleotide **G** of the codon number 17 is replaced by **A**
The nucleotide **G** number 35 is eliminated
The second nucleotide **G** of the codon number 17 is eliminated
The nucleotide **G** is added between the nucleotides number 35 and 36
The nucleotide **G** is added between the second and the third nucleotides of the codon number 17
The nucleotide **G** is added between the codons number 17 and 18

Possible effects of genetic mutations:

- Silent: no effect on the obtained peptide (or protein)
- Missense: replacement of amino acid in the peptide sequence
- Nonsense: incomplete peptide (or protein) due to the formation of stop codon
- Frame-shift: change in all the amino acids of the peptide (or protein) after the site of mutation in case of an insertion or a deletion

Ch.3 Act.4 Detection of genetic polymorphism

Restriction enzyme:

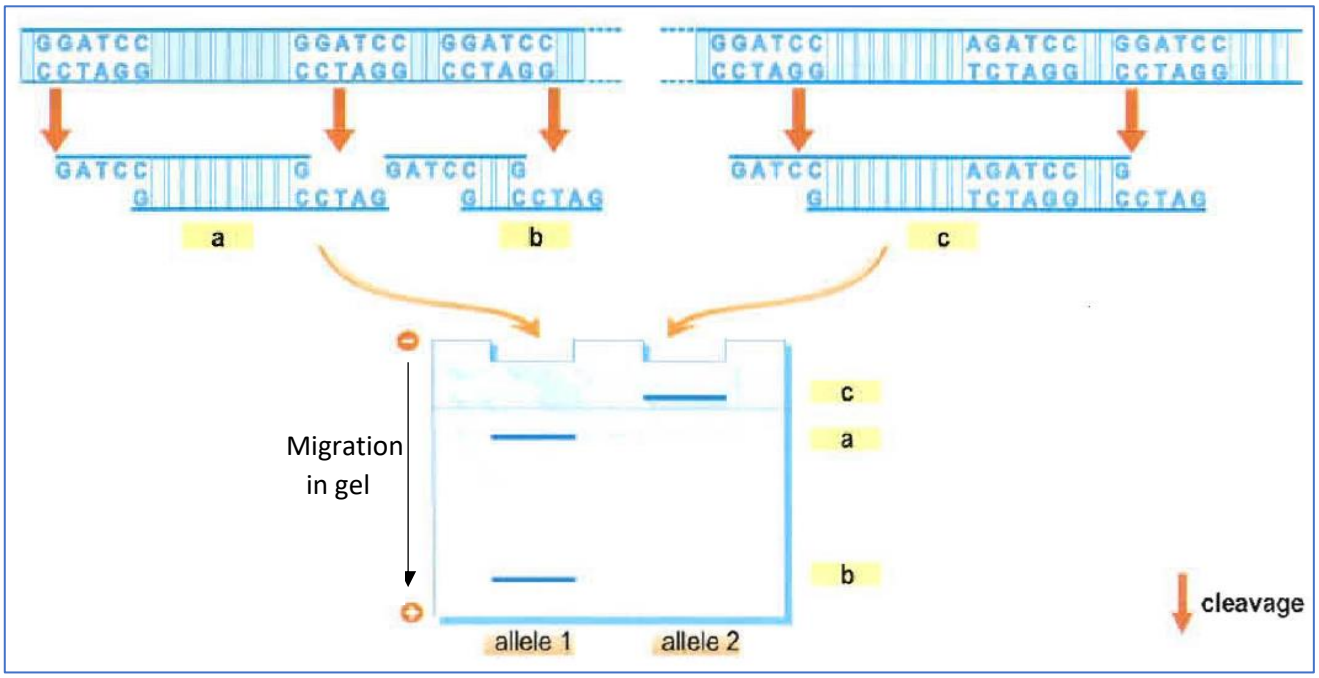
Enzyme that cuts the DNA molecule at the level of a specific sequence called recognition site (or restriction site). For example, the enzyme EcoRI cuts at the level of the following nucleotide sequence: 

If the restriction enzyme cuts the DNA *(n)* times, we obtain *(n+1)* fragments.

If the restriction enzyme cuts a gene in the DNA *(n)* times, we obtain *(n-1)* fragments. The two other fragments on both sides of the gene remain in the well because they are very big to pass in the gel of the electrophoresis.

Detection of genetic polymorphism by electrophoresis:

If a mutation occurs within the recognition site, the restriction enzyme will not recognize the sequence and hence cuts two different alleles of the same gene in different ways.



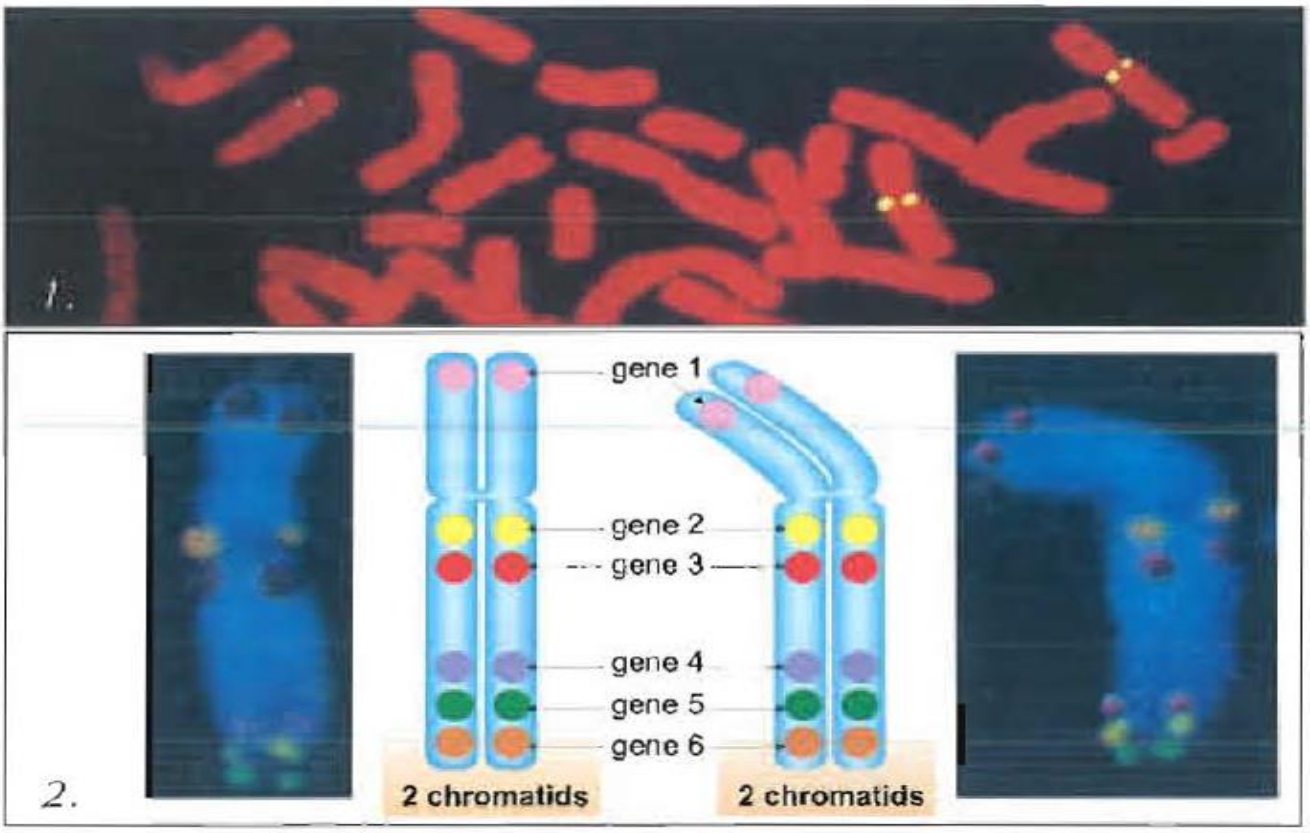
Restriction Fragment Length Polymorphism (RFLP):

Mutations occurring in non-coding regions do not usually affect the phenotype, and hence can only be revealed by variations in restriction maps of the regions in question. This is because the restriction map is independent of gene function. A difference in restriction maps between two individuals is called a restriction fragment length polymorphism or RFLP

Ch.3 Act.5 Genetic identity of individuals

FISH technique:

Technique used to **localize a gene** on a chromosome using **fluorescent** probe (small DNA sequence compementary to the studed gene) in order to produce a genetic map.



DNA fingerprint (Southern blot or Jeffrey's technique):

Technique used to **produce a restriction map** using **radioactive** probe in order to make a paternity test, detect a criminal and other applications in the legal domaine