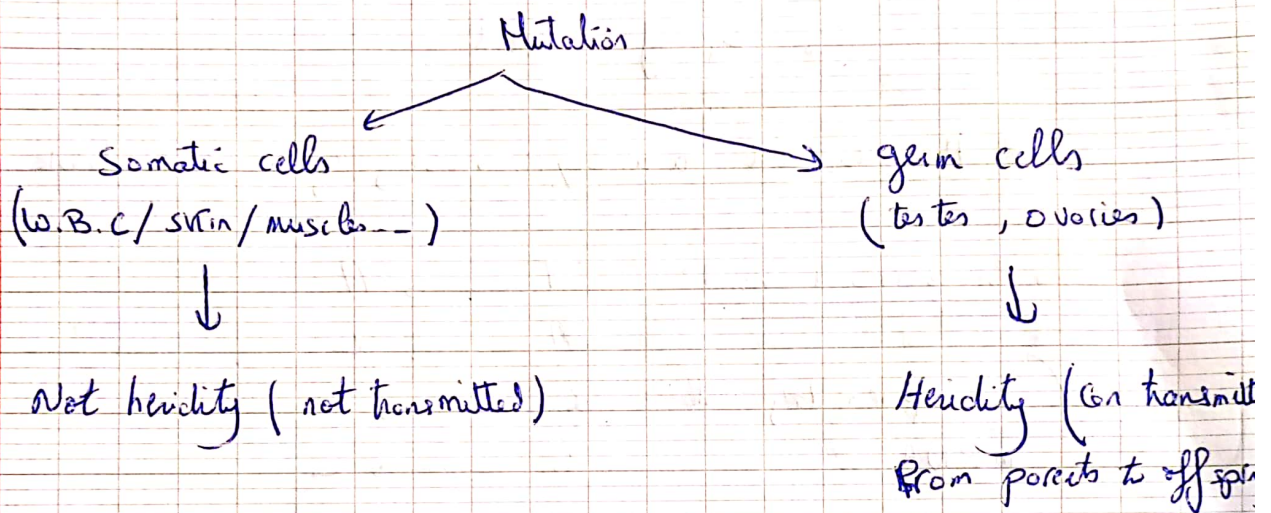


## Chapter 3

### → Doc 1: Mutation & environment:

\* Mutation is any change in nucleotide seq of certain gene.

\* How mutation can be transmitted:



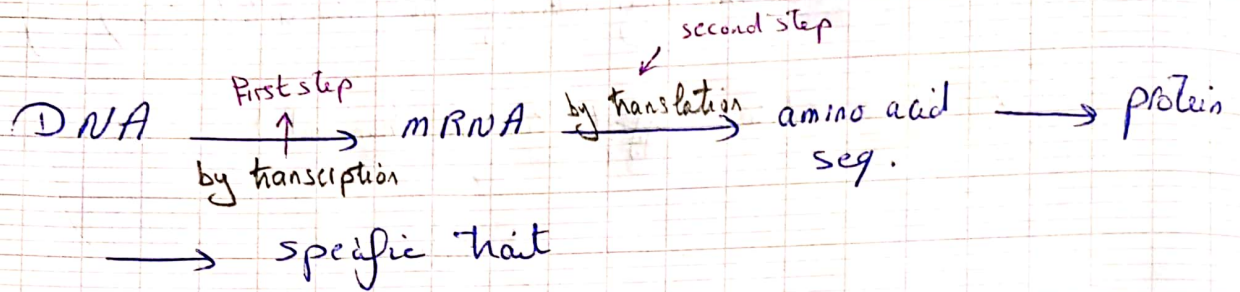
\* How environment affect Mutation:

environment agents caused by some mutation → gene coding for protein → that leads to abnormal protein →  
↑ growth → skin cancer.



## → Doc. 3: Mutation & multiple alleles:

### protein synthesis:



### \* How to det. a.a. seq.: using genetic code table

#### • transcribed strand:

→ Complementary with mRNA

T  $\rightarrow$  A  
A  $\rightarrow$  U  
C  $\rightarrow$  G  
G  $\rightarrow$  C

#### • Non-transcribed strand:

→ identical to mRNA

T  $\rightarrow$  U

• Note: Codon. (AUG --)  
                                  ↑  
                                  1<sup>st</sup> nucleotide

#### • Notes

any change DNA seq. may lead to change a.a. seq  $\rightarrow$  change phenotype (trait).



## \* Types of Mutation:

① substitution: replaced nucleotide by another one.

types for substitution:

- Silent: it change the DNA and mRNA only  
- a.a. seq doesn't change or doesn't affect

- non-sense - In a.a. → stop Codon

thus translation become incomplete.

- Missense → change 1 nucleotide → change mRNA  
→ change a.a. seq. (Rep. 1 DNA by another) or more

ex.ii

A C C ← DNA

U G G ← mRNA

Trp ← a.a.

U C C

A G G

trans.

Arg

A replaced by U

② Deletion: one nucleotide seq. disappears  
also a short seq. B.L. ←

ex.ii

TAC - ACC - GAT - A...

UAC - ACC - GAU - A...

(Y) - Thr - Asp.

(1 or many nuc. are lost)

TAC - CCG - ATA

UAC - CCG - AUA

(Y) - pro - Ile

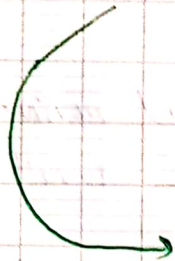
Non sense

③ Insertion: 1 or more nucleotide was added

Non-frames:

ex:

TAC - ACC - ACG - A ---  
UAC - ACC - ACG - A ---  
Tyr - Thr - Thr



TAC - GAC - CAC - GA ---  
Thy - Asp - His

was added

\* Mutation in Blood groups

- Blood A → antigen A on surface
- Blood B → antigen B " "
- Blood O → antigen O

The 3 Blood groups common in (H antigen), but differ in sugar added.

\* Genetic polymorphism is due to mutations affecting the DNA seq of genes.



## \* How to answer Q Compare:

\* Compare → total n.b of nucleotide seq. ( $>$  or  $<$ )  
Both nucleotide seq are identical except in ( — 8 — ) triplets where — present in normal but absent in mutated.

For  
deletion  
and insertion

\* Compare → number of nucleotide seq of both alleles or genes identical except in — nucleotide of — triplet where — rep. by —

For  
substitution

(imp.) \* Q: explain how change n. seq leads — 8

Mutation by — in gene coding for — in — and — triplet  
⇒ leads to (change longer shorter) mRNA by transcription  
⇒ leads to change a.a seq by translation  
⇒ change structure of —  
⇒ abnormal —  
⇒ + treat

## → Doc. 4 : Detection of genetic polymorphism

\* Restriction enzymes → cut the DNA molecule at specific site.

• n.b of restriction fragments = n.b of cleavage site + 1

• Size of restriction fragments:

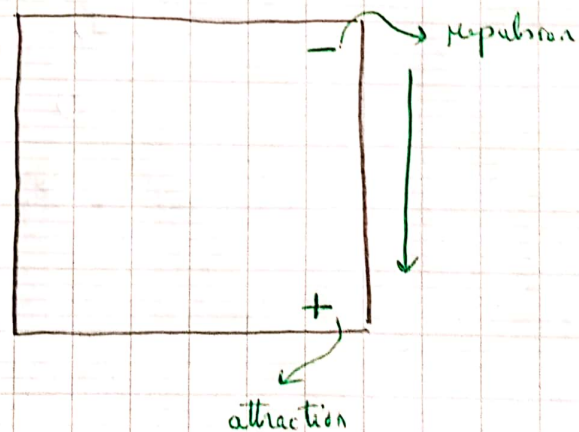
- In normal allele cuts bet — in position — and — in position — that give fragment of length —  
↑  
الأليل - الأليل

\* Gel electrophoresis:

↳ technique used to sep. DNA fragments acc. to their size.

→ large fragments migrate slower than small fragments

→ DNA migrated to the pole (from -ve pole to +ve)





## → Doc 5: Genetic Identity

### \* FISH Method

- ↳ steps:
- Denaturation: 1 sep of 2 strands of DNA
  - Hybridization: add a monoclous probe that binds to specific Complementary DNA
  - Visualization:

↳ Aim: to determine the locus of gene on Metaphase.

\* Genetic Map: it is constructed by using  $\neq$  monoclous probe with diff. colors to locate many genes.

### \* DNA Fingerprint

- ↳ steps:
- 1 - Cut the double strand DNA with specific restriction enzyme
  - 2 - Electrophoresis
  - 3 - Southern Blotting
  - 4 - Denaturation & Hybridization
  - 5 - Autoradiography

# DNA Analysis

