

血 Hematology 血



Histology

Biochemistry

Pathology

Pharmacology

Physiology

Microbiology

Handout

Slide 7

Sheet



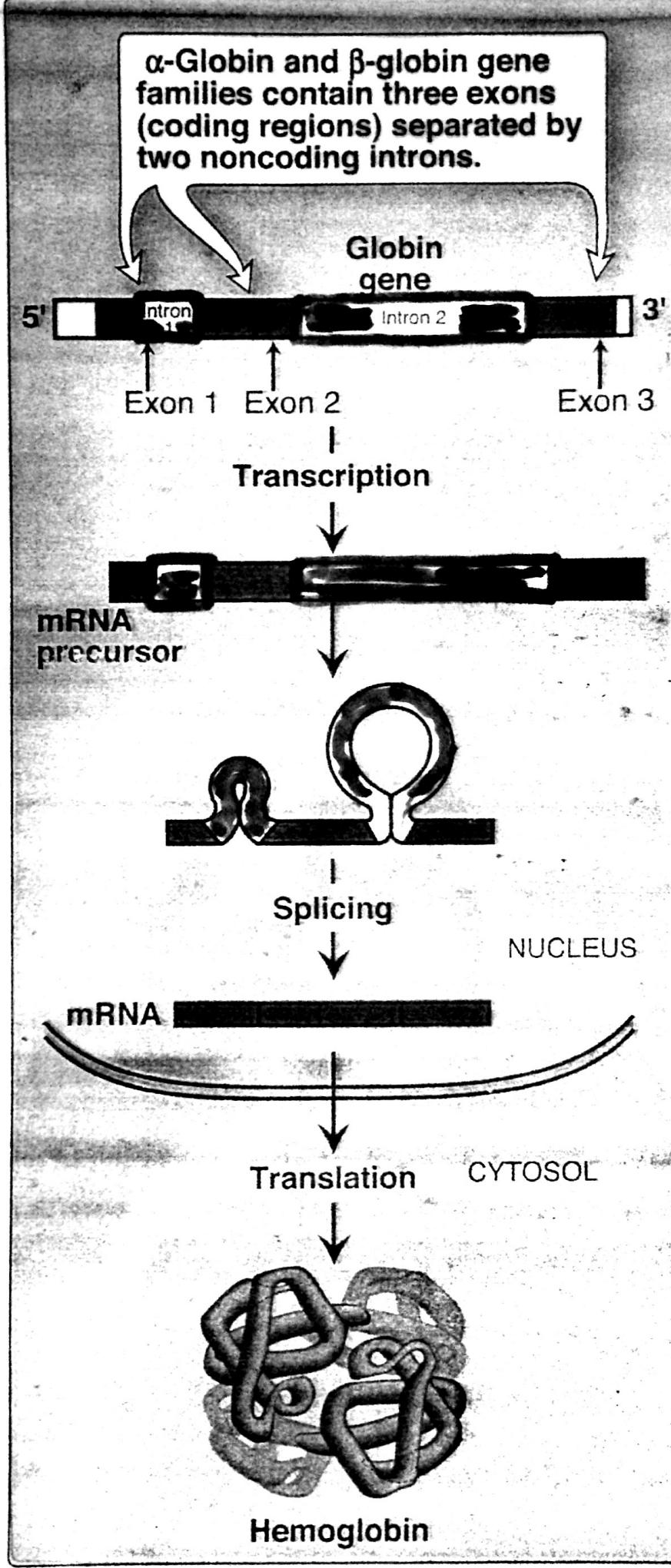
Dr. name :
Dr Nayef karadsheh

Lecture number :

Done BY :



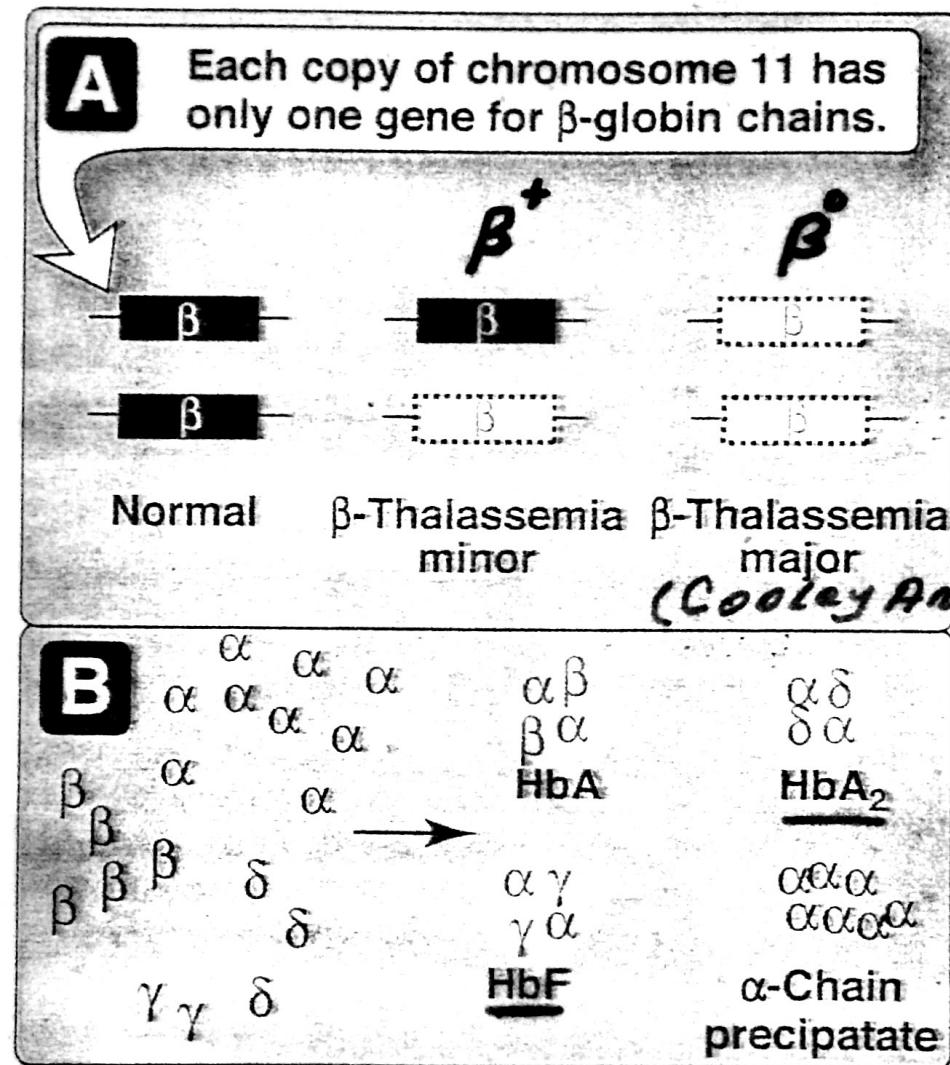
Globin chains Synthesis



THALASSEMIA

- Decreased Synthesis of α - or β -
- Globin chain precipitate
- Hemolysis
- Hypochromic anemia

β -thalassemia



$HbA_2 \uparrow$; $HbF \uparrow$

α chains $\rightarrow \alpha_4$ (Cooley's Hb)

→ α_4 precipitates

→ Heinz bodies

→ Cell membrane damage → Premature death of erythrocyte

Manifestation of β -thalassemia appears only after birth (because of HbF); becomes severely anemic during the 1st and 2nd year of life.

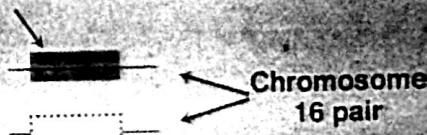
α -THALASSEMIA

13b

A

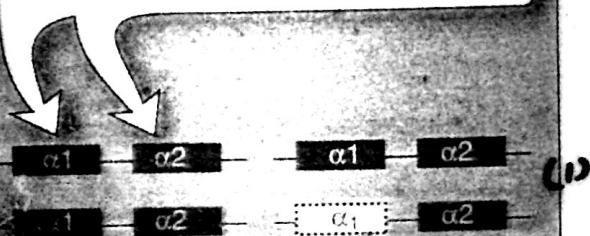
Key to symbols

Normal gene for
 α -globin chain



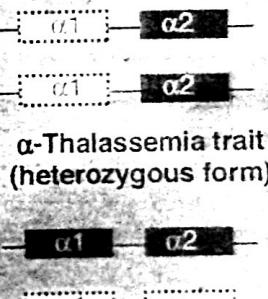
Deleted gene for
 α -globin chain

Each copy of chromosome 16 has
two adjacent genes for α -globin chains.



Normal
individuals

"Silent"
carrier



α -Thalassemia trait
(heterozygous form)

α -Thalassemia trait
(heterozygous form)

(2)
Show some
mild symptoms
clinically

(3)
Hemoglobin H
disease
(variable severity)

Hemoglobin Bart
disease with hydrops
fetalis (usually fatal
at birth) or before

in total loss of
 α -chain

B

α α

$\alpha\beta$
 $\beta\alpha$

HbA

$\beta\beta$

$\beta\beta$

$\beta\beta$

HbH₂
(precipitates
forming
Heinz
bodies)

Hb Bart

74

HbH ↑
Hb Bart ↑

Soluble Hb
but without sigmoidal
kinetics. Useless O₂
deliverer to tissues.

Primary Causes :

α - thalassemia :-
gene deletion

β - thalassemia :-

- Point mutation in the Promotor
- mutation in the translational initiation codon
- point mutation in the Polyadenylation signal
- mutations \rightarrow splicing abnormalities

- $HbE \beta^{26\text{Glu-Lys}}$
structural and quantity abnormality
60% of β -globin is made

- Hereditary persistence of fetal Hb (HPFH)

- \rightarrow continue to make HbF in adult
- \rightarrow Benign