CASE4:
Pale and tired

PBL
TEAMWORK

❖Learning issues:

- ✓ Structure and function of RBCs
- ✓ Structure and functions of normal hemoglobin (HbA2). Other types of hemoglobin and physiological differences
- ✓ Development of RBCs and physiological factors needed for normal development in the bone marrow
- ✓ Physiology and metabolism of iron
- ✓ Mechanisms by which anemia may develop
- ✓ Pathogenesis of beta thalassaemia major. Differences between major and intermedia
- ✓ Interpretation of the patient symptoms, signs and investigation results
- ✓ A brief management plan showing management goals and management options

- MALE
- 7 years old
- Always tired *
- Pale *
- Does not participate in any physical education classes
- Used to be active
- Short of breath *
- Declined activity over the last (10-12) months

HISTORY:

- No bronchial asthema
- No history of blood loss
- No blood transfusion
- No previous hospital admission
- Born in small village in Jeezan
- His mother was not seen by a doctor during pregnancy
- No allergies
- Decline in his school preformance in most subjects

Family History: His half-brother is always ill and needs blood transfusion nearly every month. (it's usually associated with hemolysis).

(*) presenting problems

Examination:

* Pale, no skeletal mucsle problems, vital signs are normal EXCEPT for increased pulse rate (TACHYCARDIA)
* NORMAL CVS , NORMAL RESPIRATORY SYSTEM
* NORMAL ABDOMINAL EXAMINATION

Blood tests :

Blood test	Aymen's results	Normal range
HB	78	130-170 g/L
MCV	78	83-101 fL
MCH	26	27-32 pg
Platelets count	200	150-400 x 10^9/L

Because of the low MCV , MCH , and presence of microcytic hypochromic RBCs Doctor :decided to do further tests to confirm Iron-deficiency anemia

Blood test	Aymen's results	Normal range
Serum iron	31	9-30 µmol/L
Serum ferritin	120	10-120 mg/L
Serum transferrin	2.5	2.0-4.0 g/L

Blood film report:

Aymen's blood film shows:

- Hypochromasia (RBCs are paler than normal cells)
-Microcytosis (RBCs are smaller than normal cells)
-Polychromasia (RBCs tend to be stained with acid and basic days)
-Target cells (RBCs with a dark center surrounded by a light band that again is encircled by a darker ring)
-Anisocytosis (significant variations in the size of RBCs)

Doctor arrange for **(haemoglobin electrophoresis)** to identifying the exact cause of the anemia

Haemoglobin type	Aymen's result	% of total haemoglobin
Hb A	52%	Over 95%
Hb A2	7%	2-3%
Hb F	41%	Less than 1%
Hb S	Absent	Absent
Other abnormal Hb	Absent	Absent

hemoglobin electrophoresis test is a blood test done to evaluate the different types of hemoglobin in the bloodstream

Diagnosis :

Beta Thalassaemia intermedia (Anemia due to an inherited blood condition)

Intermedia because :

1-Hb in the range of 7.5-8.5 g/dL without the need for blood transfusion
2-Presented at relatively later age (7 years old)
3-Blood changes are not severe → he will not need blood transfusion **except on the presence of severe infection or hyper active spleen destroying the RBCs**

Structure and function of RBC

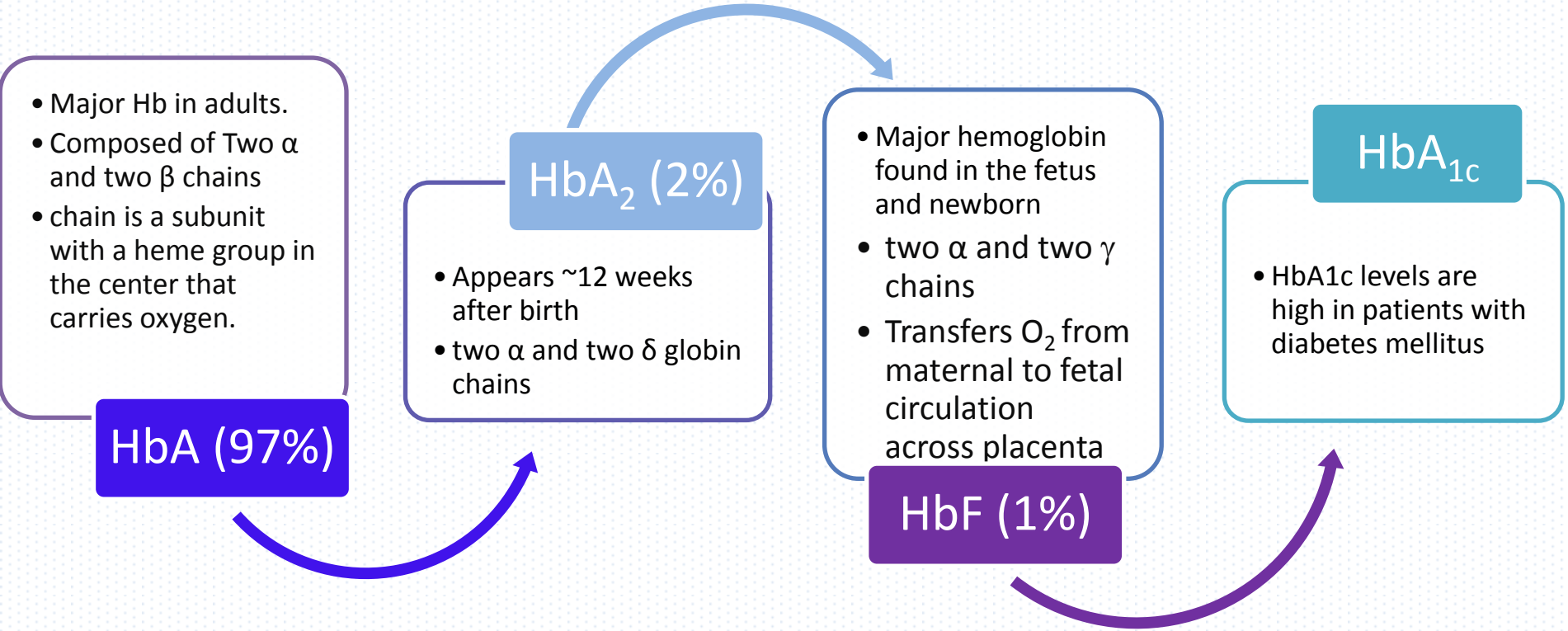
-small and biconcave in shape

-they lack a nucleus

-contain hemoglobin

The most important function of red blood cells is the transport of oxygen.

Structure of hemoglobin



Thalassemia

β-Thalassemia

The condition in involving β-thalassemia is one of 2 categories : (molecular classification)
Beta0: the synthesis of Beta globin is completely lost.
Beta+: the synthesis of Beta globin is partially decreased.

α-Thalassemia

Deletion involving one or more of α-globin genes.

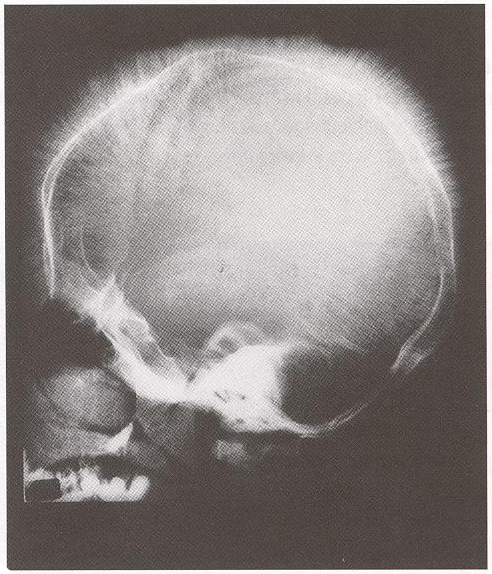
Clinical classification of β-Thalassemia

Type	Clinical features
β-Thalassemia major	Severe anemia Regular blood transfusion required
β-Thalassemia intermedia	Severe anemia Regular blood transfusion is not required
β-Thalassemia minor	Asymptomatic, mild or absent anemia

The onset of thalassemia major is at early life nearly at birth while thalassemia intermedia appears at later age (As in the case)

Signs and symptoms

*Pallor
*Jaundice (because of hemolysis)
*Apathy and Anorexia
*Failure to Thrive
*Hepato-splenomegaly
*Skeletal Deformity (Hair end appearance under x-ray)
*Iron Overload mainfestations.



hair-like projection (as a result of skeletal deformity)

Hemoglobin electrophoresis for Beta thalassemia:

B-thalassemia type	HbA	HbA2	HbF
major	0	1.5-5.9	More than 94
Intermedia	Present	5.4-10	30-73
Minor	More than 90	3.5-8	1-2

Questions

Q1: what's ayman presenting problems?	**Q2: what is the diagnose in ayman case and what is the causes of it ?**
(tiredness – pallor - decrease in physical activity)	He have thalassemia (microcytic hypochromic animia), due to an inherited blood condition
Q3: why he has shortness of breath and tachycardia?	**Q4: What are the tests you will order in this case?**
because RBC's are not normal so less oxygen will be carried to tissues leading to increase heart rate to compensate the oxygen loss.	-Complete blood count, hemoglobin electrophoresis, blood film -Serum transferrin, ferritin, Fe Binding Capacity (Iron studies to exclude iron deficiency anemia)
Q5: 'target cell' is an important finding in aymn blood film , why?	**Q6: Why CBC is not enough in this case ?**
It's indicate thalassaemia	Because iron deficiency anemia and thalassemia are similar to each other.
Q7: Tiredness, pallor and shortness of breath are signs for what ?	**Q8: What does anemia mean ?**
Anemia	Mean less than normal hemoglobin and RBC's are unable to carry oxygen in a way to cover all body need's.

Q9: What is the goal of long-term blood transfusion?	**Q10: in this case, why he is not need the blood transfusion ?**
To maintain the patient's hemoglobin level at 9-10 g/dL.	because it is intermedia (Hb in the range of 7.5-8.5 g/dL) and Blood changes are not severe
Q11: what is the difference between Iron-deficiency anemia and Thalassaemia ?	**Q12: after Exclude the Iron-deficiency anemia , What is the test that doctor should decided to do to identifying the cause of the anemia?**
Thalassemia : an inherited disorder caused by mutation that decrease the synthesis of α or β-globin chains (completely or partially missed) **Iron-deficiency anemia :** most common , with low Serum iron , low Serum ferritin And low Serum transferrin **Both are microcytic hypochromic anemia type**	hemoglobin electrophoresis test
Q13: What is hemoglobin electrophoresis ?	**Q14: what is the Clinical Manifestations in this case ?**
test is a blood test done to evaluate the different types of hemoglobin in the bloodstream	Pallor , jaundice , Hepato-splenomegaly, Skeletal Deformity, Iron Overload mainfestations

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