

1st course.pathology

RBC.DISORDES



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. Third Stage

المرحلة الثالثة

الجانب النظري محاضرة رقم 12

Lecture No. 12 Theoretical

اتدريسي المادة : م.م.فاضل عوفي عواد



Red Blood Cell Disorders



- Definition: RBCs carry oxygen from lungs to tissues and CO₂ from tissues to lungs.
- Functions:
 - 1. Oxygen transport via hemoglobin.
 - 2. Acid-base balance support.
 - 3. Maintain blood viscosity.
- RBC Disorders: Include underproduction, destruction, and genetic abnormalities.



Classification of RBC Disorders



- 1. Hypoproliferative Anemia (Underproduction)
 - - Caused by iron, vitamin B12, or folate deficiency.
- 2. Hemolytic Anemia (Destruction of RBCs)
 - - RBC breakdown in blood vessels or spleen.
- 3. Genetic RBC Disorders
 - - Thalassemia, Sickle cell anemia, Sideroblastic anemia.
- 4. Macrocytic and Non-Macrocytic Anemia

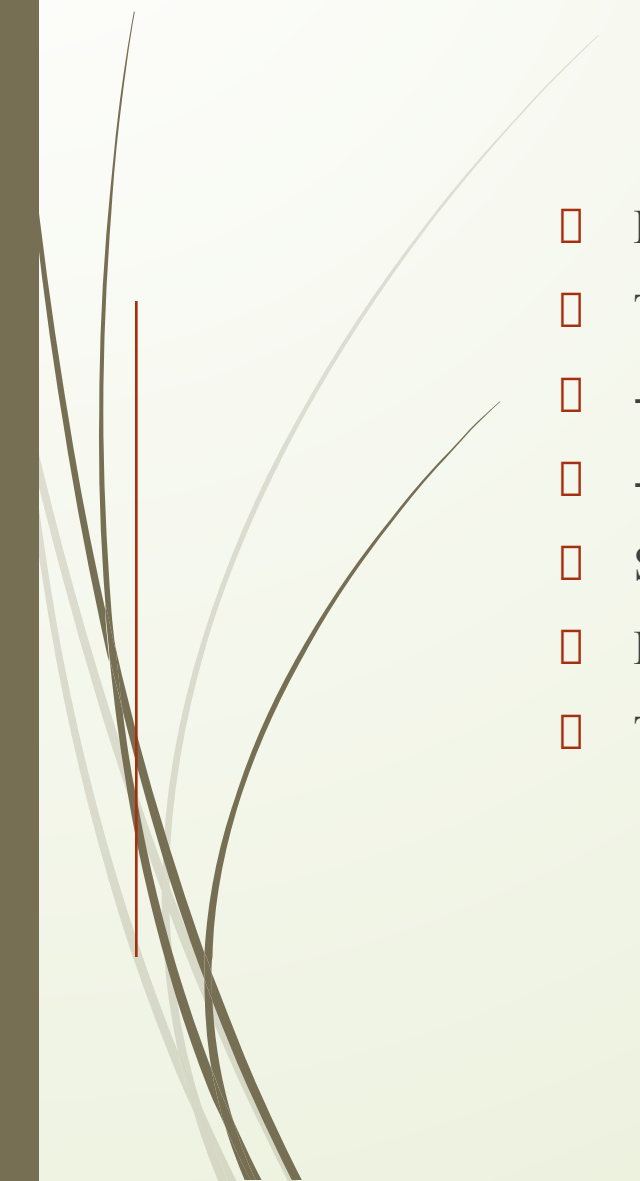


Sideroblastic Anemia

- Definition: RBCs cannot incorporate iron into hemoglobin → iron accumulates in mitochondria.
- Causes:
 - - Inherited: ALAS2 gene mutation
 - - Acquired: Drugs (isoniazid, chloramphenicol), B6 deficiency
- Symptoms: Fatigue, pallor, hemolysis, iron overload
- Diagnosis: CBC: microcytosis, low Hb; Bone marrow: Ring sideroblasts
- Treatment: Vitamin B6, transfusions if needed



Thalassemia



- Definition: Genetic disorder → reduced or absent hemoglobin chain synthesis (α or β).
- Types:
 - - α -Thalassemia: α -chain deficiency
 - - β -Thalassemia: β -chain deficiency
- Symptoms: Chronic anemia, splenomegaly, growth delay
- Diagnosis: CBC: microcytic hypochromic anemia; Hemoglobin electrophoresis
- Treatment: Regular transfusions, bone marrow transplant in severe cases



Macrocytic Anemia



- ❑ Definition: Enlarged RBCs (MCV >100 fl)
- ❑ Causes: Vitamin B12 deficiency (pernicious anemia), Folate deficiency, Drugs: methotrexate, chemotherapy
- ❑ Symptoms: Weakness, pallor, neurological deficits (B12 deficiency)
- ❑ Diagnosis: CBC: high MCV, B12 and folate levels
- ❑ Treatment: B12 or folate supplementation



Non-Macrocytic Anemia



- Definition: RBCs normal or small (MCV 80–100 fl or <80 fl)
- Causes: Iron deficiency anemia, Anemia of chronic disease
- Symptoms: Fatigue, pallor, shortness of breath
- Diagnosis: CBC, iron studies, ferritin
- Treatment: Iron supplementation, treat underlying cause



Anemia Due to Underproduction

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- ❑ Causes: Bone marrow failure (aplastic anemia), Chronic kidney disease → low erythropoietin, Endocrine disorders
 - ❑ Symptoms: Fatigue, pallor, infections (if aplastic anemia)
 - ❑ Diagnosis: CBC, reticulocyte count, bone marrow biopsy
 - ❑ Treatment: Treat underlying cause, erythropoietin, transfusions



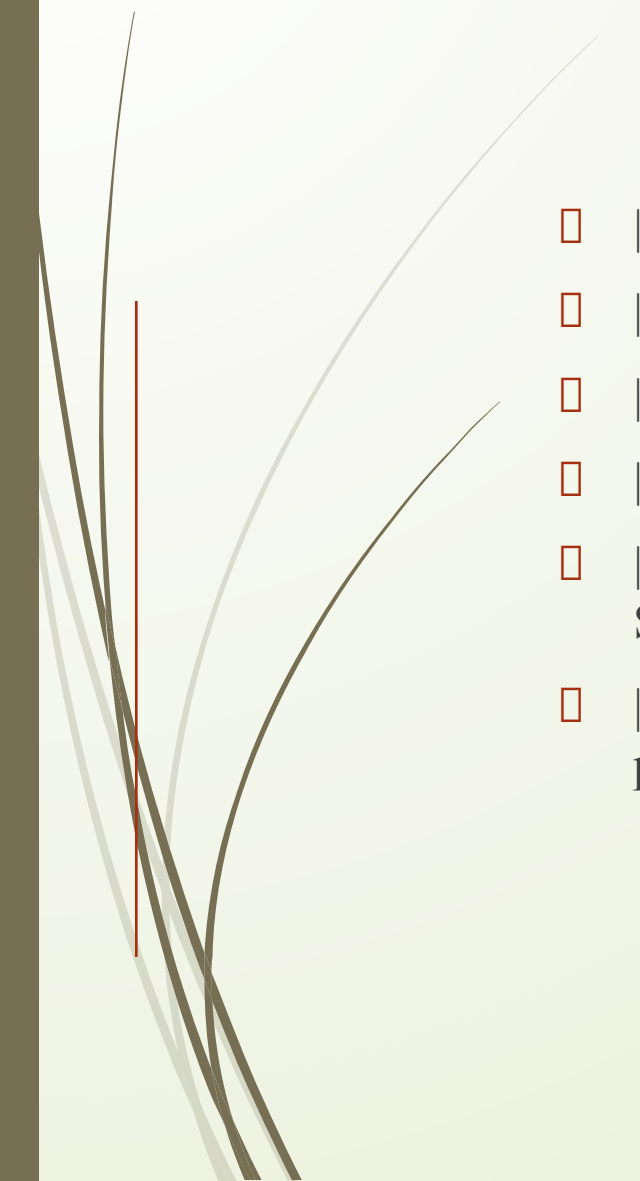
Hemolytic Anemia



- Definition: Premature destruction of RBCs
- Causes: Intrinsic defects: membrane, enzyme, hemoglobinopathies; Extrinsic: autoimmune, infections, drugs
- Symptoms: Jaundice, pallor, dark urine, splenomegaly
- Diagnosis: Elevated LDH, bilirubin, reticulocytes
- Treatment: Treat cause, immunosuppressants if autoimmune, transfusions if severe



Summary Table



□	Disorder RBC Size Cause Key Feature Treatment
□	----- ----- ----- ----- -----
□	Sideroblastic Microcytic Iron incorporation defect Ring sideroblasts B6, transfusions
□	Thalassemia Microcytic Genetic Hb chain defect Chronic anemia Transfusion, BMT
□	Macrocytic Anemia Macrocytic B12/Folate deficiency Neuropathy (B12) Supplements
□	Non-Macrocytic Normocytic/Microcytic Iron deficiency / chronic disease Fatigue, pallor Iron / treat cause



Key Points & Take-Home Messages



- - RBC disorders can be genetic, nutritional, or acquired.
- - Diagnosis relies on CBC, iron studies, B12/folate, bone marrow, hemoglobin electrophoresis.
- - Treatment is cause-specific: supplements, transfusions, or bone marrow transplant.
- - Early detection prevents complications like growth delay, organ damage, and neurological deficits.

Any questions?

*Thank
You*