1st course.pathology

RBC.DISORDES

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الجانب النظري محاضرة رقم 12

Theoretical Lecture No. 12

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Red Blood Cell Disorders

- Definition: RBCs carry oxygen from lungs to tissues and CO_2 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

- 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?