Lec 10: Polycythemia

- Polycythemia refers to an increased concentration of red blood cells (RBCs) in the blood, resulting in elevated hemoglobin levels, hematocrit, and RBC count.
- This condition thickens the blood and can lead to complications such as clotting, stroke, or heart strain.

Types of Polycythemia

Polycythemia refers to an increased concentration of red blood cells in the blood. It is classified into three main categories based on the underlying cause:

Polycythemia Classifications

1. Primary Polycythemia

• Cause: Autonomous proliferation of red blood cells in the bone marrow.

• Key Example: Polycythemia Vera (PV)

• A myeloproliferative neoplasm caused by mutations (e.g., JAK2 mutation).

• Associated with low erythropoietin (EPO) levels.

• Often involves increased production of white blood cells and platelets.

2. Secondary Polycythemia

• Cause: Increased erythropoietin (EPO) production due to hypoxia or other stimuli.

•**Causes Include:** 1.Chronic Hypoxia, 2.Chronic obstructive pulmonary disease (COPD), 3.High-altitude living., 4.Obstructive sleep apnea.,5.EPO-Secreting Tumors, 6.Renal cell carcinoma,7.Hepatocellular carcinoma., 8.Use of anabolic steroids or erythropoietin injections.

3. Relative Polycythemia

A decrease in plasma volume, leading to a relative increase in red blood cell concentration. Common Causes:

1.Dehydration (e.g., from diarrhea, vomiting, or inadequate fluid intake).

2.Gaisböck syndrome (stress erythrocytosis in overweight, hypertensive individuals).

Summary Table

Type	Cause	Epo level	Example
Primary Polycythemia	Bone marrow proliferation	low	Polycythemia vera(PV)
Secondary Polycythemia	Increased Epo(due to hypoxia or tumer)	high	COPD high altitude, tumers
Relative Polycythemia	Decreased plasma volume	Normal or low	Dehydration, gaisbock syndrome

Clinical Features

• Symptoms :Headache, dizziness, blurred vision , Fatigue, itching (especially after a hot bath in PV) • Red or flushed skin, especially in the face.

• Complications:

• Thrombosis (e.g., deep vein thrombosis, stroke).

• Bleeding (due to abnormal platelet function in PV).

Diagnosis of Polycythemia:

Diagnosing polycythemia involves clinical evaluation, laboratory tests, and imaging to determine the cause and type (primary, secondary, or relative).

A. Clinical Evaluation

• Symptoms: Headache, dizziness, fatigue, flushing, itching (especially after a hot bath in Polycythemia Vera), or thrombosis.

• History :Smoking, living at high altitudes, or chronic respiratory diseases (secondary causes), Medications like erythropoietin or anabolic steroids , Family history of myeloproliferative disorders.

B. Laboratory Tests

1. Complete Blood Count (CBC):

• Elevated hemoglobin and hematocrit: Hb 16.5 g/dL (men), >16 g/dL (women) , Hematocrit >49% (men), >48% (women).

2. Blood film Smear: May show increased red blood cells and occasionally immature granulocytes in Polycythemia Vera.

3. Serum Erythropoietin (EPO) Level:

• Low EPO: Suggests Polycythemia Vera (primary).

• High EPO: Indicates secondary polycythemia (e.g., hypoxia, EPO-secreting tumors.(

4. Oxygen Saturation and Arterial Blood Gases:

• Assess hypoxia as a cause of secondary polycythemia.

5. JAK2 Mutation Testing:

• Positive in >95% of cases of Polycythemia Vera.

6. Imaging and Additional Tests

• Chest X-ray/CT Scan: To evaluate lung diseases causing hypoxia.

• Abdominal Ultrasound/CT: Detect tumors (e.g., renal or hepatic carcinomas).

7. Bone Marrow Biopsy:

• Hypercellularity confirms Polycythemia Vera.

Differentiating Between Types

feature	Primary pv	Secondary	Relative
EPO level	Low	High	Normal /low
JAK2 Mutation	Positive	Negative	Negative
Oxygen saturation	Normal	Decreased	Normal
Plasma volume	Normal	Normal	Reduced

Diagnostic Criteria for Polycythemia Vera:

Requires all 3 major criteria or the first 2 major criteria and 1 minor criterion:

Major Criteria:

1. Elevated hemoglobin (>16.5 g/dL in men; >16 g/dL in women) or hematocrit (>49% in men; >48% in women).
2. Bone marrow biopsy showing hypercellularity.
3. JAK2 mutation (V617F or exon 12).

Minor Criteria:

• Low serum erythropoietin level.

Summary Approach

1. Start with CBC to confirm elevated RBC levels.
2. Measure serum EPO and check JAK2 mutation status.
3. Rule out secondary causes with imaging, oxygen assessment, and clinical history.
4. If needed, perform bone marrow biopsy for definitive diagnosis.

Management

• Primary Polycythemia (e.g., PV):

• Phlebotomy: To reduce hematocrit (<45%.(

• Medications: Hydroxyurea (to reduce cell production), low-dose aspirin (to prevent thrombosis).

• Targeted Therapy: JAK2 inhibitors (e.g., ruxolitinib.(

• Secondary Polycythemia:

• Address the underlying cause (e.g., oxygen therapy for hypoxia, treating tumors).

• Relative Polycythemia:

• Correct dehydration or manage stress.

Prognosis

With proper treatment, complications can be minimized, especially in primary polycythemia like PV. Left untreated, the condition increases the risk of serious vascular events.

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