Polycythemia

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DEFINITION

- Increase in total RBC mass above normal range
- Erythrocytosis: increased RBC s number
- Relative polycythemia: secondary to decreased plasma volume (water deprivation, severe diarrhea, diuretics)

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ABSOLUTE POLYCYTHEMIA

- Absolute polycythemia: true increase in RBC mass, secondary to increased bone marrow (BM) production
- Can be primary or secondary
- Primary: autonomous high bone marrow production (polycythemia vera), erythropoietin is low
- Secondary: systemic hypoxia → high erythropoietin → increased erythropoiesis

SECONDARY POLYCYTHEMIA

- Adaptive: living in high altitude, cyanotic heart disease, chronic pulmonary diseases, sleep apnea
- Paraneoplastic: renal cancer, liver cancer
- Surreptitious (blood doping): endurance athletes
- Alcohol: frequent urination, depressed respiration (both relative and absolute)
- Smoking
- In secondary polycythemia: high erythropoietin, no splenomegaly



POLYCYTHEMIA VERA

- Myeloproliferative neoplasm
- Mutation in tyrosine kinase JAK2 in bone marrow stem cells
- Normally acts in the signaling pathway of erythropoietin receptor and other growth factor receptors
- Hematopoietic cells become less dependent on growth factors (negative feedback → low erythropoietin level)
- Excessive proliferation of erythroid, myeloid cells and megakaryocytes (panmyelosis)
- Erythrocytosis is most prominent, results in polycythemia
- Splenomegaly is common



SYMPTOMS OF POLYCYTHEMIA

- Plethora (red skin) / cyanosis (blue)
- Headache and dizziness (from hypertension)
- Slow circulation and hyperviscosity cause cyanosis, blurred vision, tissue ischemia
- Thrombosis, or bleeding (disturbed function of vWF)

In polycythemia vera: similar symptoms plus:

- Pruritus (aquagenic)
- Peptic ulcer
- Secondary gout (arthritis, kidney stones, tophi)
- Chronic disease
- Spent phase: occurs after an interval of 10 years of symptoms, BM becomes fibrotic, hematopoiesis shifts to spleen
- Blast crisis: transformation to acute myeloid leukemia (rare)



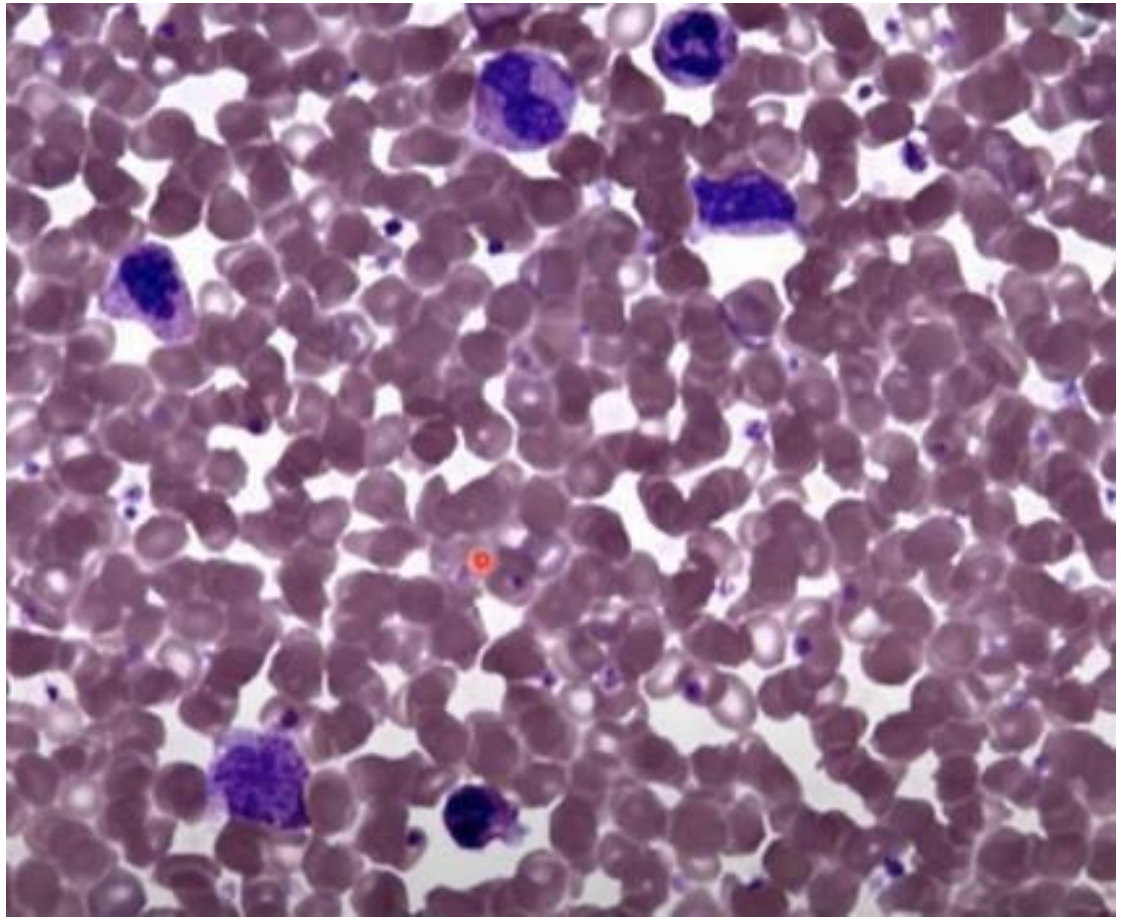
LABORATORY FINDINGS OF POLYCYTHEMIA

- High hemoglobin concentration (>16.5 g/dL in men, 16 in women) and high hematocrit (>49% in men, 48% in women)
- High RBCs count
- These tests might be masked (appear normal) if iron deficiency develops

In polycythemia vera: additional findings:

- Leukocytosis and thrombocytosis are common
- JAK2 gene mutation: positive
- Low erythropoietin level
- Hypercellular bone marrow with panmyelosis





- Peripheral blood smear in polycythemia: packed RBCs