

# Polycythemia

## Symptoms

- In General**
  - \*Plethora/cyanosis
  - Headache and dizziness (from hypertension)
  - /Slow circulation and hyperviscosity cause cyanosis, blurred vision, tissue ischemia
  - Thrombosis, or bleeding (disturbed function of vWF)
  - Pruritus (aquagenic)
  - Peptic ulcer
  - secondary gout (arthritis, kidney stones, tophi)
- In polycythemia vera: similar symptoms plus**
  - 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)

## Definitions

- Polycythemia : Increase in total RBC mass above normal range
  - Erythrocytosis: increased RBCs number
  - Relative polycythemia: secondary to decreased plasma volume (water deprivation, severe diarrhea, diuretics)
  - Primary: polycythemia vera (low erythropoietin, splenomegaly)
  - Absolute polycythemia: true increase in RBC mass, secondary to increased BM production

## Tow types of polycythemia

### Relative polycythemia

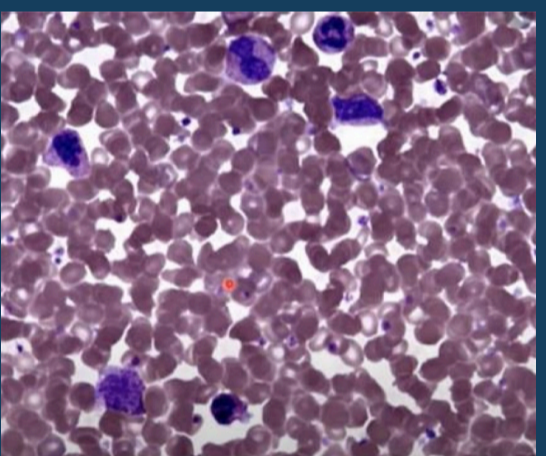
### Absolute polycythemia

**Primary (polycythemia Vera):** autonomous high bone marrow production erythropoietin is low

**Secondary polycythemia :** systemic hypoxia → high erythropoietin → increased erythropoiesis

## 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 if iron deficiency develops
- in polycythemia vera: additional findings:
  - Leukocytosis and thrombocytosis are common
  - JAK2 mutation
  - Low erythropoietin level
  - Hypercellular bone marrow with panmyelosis



• Peripheral blood smear in polycythemia: packed RBCs

" Normally acts in the signaling pathway of erythropoietin receptor and other growth factor receptors

Mutation in tyrosine kinase JAK2 in bone marrow stem cells

Myeloproliferative neoplasm

•Hematopoietic cells become less dependent on growth factors

Excessive proliferation of erythroid, myeloid cells and megakaryocytes (panmyelosis)

Splenomegaly is common

• Erythrocytosis is most prominent, results in polycythemia

### No splenomegaly

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

Smoking

### Causes