# Polycythemia

Definition: Increase in red cell count, usually with associated increase in Hb levels

Categories:

- True Polycythemia- absolute increase in red cell volume/mass with inc. PCV, Hb
- Pseudo/spurious/stress plycythemia

- Absolute polycythemia- primary/secondary
- Relative polycythemia - reduced plasma volume eg diarrhoea, vomiing, diuretics

Asolute –

- Primary – polycythemia rubra vera - myeloproliferative disease – low EP levels

Another cause – inherited EP receptor mutations

Secondary – due to increased EP levels

Secondary polycythemia

- Due to hypoxia – suggestive history, bluish cyanoisis, spleen/pruritus absent, counts mildly raised, platelets normal, BM normal, NAP score normal, oxygen sat. decreased
- Other cause – oxygen sat. normal.

Causes of secondary polycythemia

- Compensatory due to hypoxia – lung/heart disease, high altitude, sleep apnoea,
- EP secreting tumours eg RCC, HCC(paraneoplastic syndrome), Cushing's syndrome, PCKD, Hydronephrosis, Hb mutations, androgenic steroid use, heavy smoking, methemoglobenemia

# Polycythemia Rubra Vera

- Clonal myeloproliferative disorder
- Part of the CMPDs
- Increased marrow production of all cell lines, especially erythroid cells or RBCs

Pathogenesis - JAK2 mutations due to whch affected cells are growth factor independent and very little EP levels needed to stimulate them

- Age – middle aged to elderly
- Sex – M:F = 1.2 – 1
- Incidence – 1-3/1lac population per yr

Clinical features – due to raised red cell mass and Hct – raised blood volume – disturbed blood flow – stasis and deoxygenation

- Headache, dizzness, pruritus, visual problems
- Burning in skin
- Plethoric person with cyanosis, raised skin temp

- HTN, hyperuricemia with gout
- Thrombophlebitis, splenic and portal vein thrmbosis
- GI disturbances, peptic ulcers
- Strokes, MI, infarcts and CVAs
- Splenomegaly, hepatomegaly

## Investigations

- Hb– 14-28g/dl
- Tlc raised upto 50,000/cu.mm
- Esr – vey low
- Hct raised
- Red cell mass  and count– raised
- Oxygen saturation –raised
- EP levels – low
- NAP score raised

- GBP – thick film, raised RBC mass, raised TLC, basophils, platelets, giant forms

- Bone Marrow – initially hypercellular with proliferation of all cell lines,
- Later, hupocellular with fibrosis

Treatment – Phlebotomy, JAK2 inhibitors, symptomatic, hydration of patient

Prognosis – vascular complications, AML, I 2% cases, myelofibrosis

# Differential diagnosis

spurious – No raised RBCmass, no splenomegaly, EP normal, NAP score normal

no pruritus, no engorgement of vessels, BM normal, oxygen sat. normal

- Causes – excess alcohol intake, smoking, diuretics

Stress polycythemia – obese, hypertensive, anxious peopl e– Gaisbock syndrome

Secondary to hypoxia– no splenomegaly, EP raised, O2 sat. decreased , NAP score normal

# New WHO Criteria  Polycythemia vera (PV)

**Diagnosis requires the presence of both major criteria and one minor criterion**
**or the presence of the first major criterion together with two minor criteria:**

**Major criteria**

1. Hemoglobin  18.5 g/dL in men, 16.5 g/dL in women or other evidence of
increased red cell volume*

2. Presence of *JAK2* or other similar mutation

- **Minor criteria**
- 1. Bone marrow biopsy showing hypercellularity for age with trilineage growth (panmyelosis) with prominent erythroid, granulocytic, and megakaryocytic proliferation
- 2. Serum erythropoietin level below the reference range for normal
- 3. Endogenous erythroid colony formation in vitro

Familial polycythemia – younger age, few symptoms, leucocytosis/thrombocytosis absent, good prognosis