POLYCYTHEMIA

Acquired Hydropes Neoplasm arising from malignant transformation of HSC

It is characterized by An ↑ in the RBC mass usually with ↑ Hb level

- Absolute
  - Primary (low erythropoietin level)
    - Polycythemia vera.
  - Secondary (high erythropoietin level)
    - Physiological — Congenital heart
      - Lung diseases
      - Living in high Altitude
    - Pathological — Erythropoietin
      - Renal cell carcinoma
- Relative
  - Reduced plasma volume
    - Gaisbock's syndrome
    - dehydration due to vomiting, d

POLYCYTHEMIA VERA:

It is acquired clonal myeloproliferative neoplasm arising from the malignant transformation of HSC cells.

features:

→ Erythrocytosis — Accompanied by leukocytosis & thrombocytosis

→ Increased Risk of hemorrhage

→ Strongly Associated with Activating mutations in JAK2 enzyme

PATHOGENESIS:

Normal function of JAK2 → signalling pathways downstream of the erythropoietin Receptor

↓

The Activation of signalling pathway causes hematopoietic cell to Replicate

→ Acquired somatic mutation in JAK2 gene

↳ leads to dependency of HSC on growth factor

Polycythemia vera vs Relative polycythemia

Associated with → Result from hemoconcentration.

low level of serum erythropoietin & cause growth factor independent proliferation of the neoplastic clone.

CLINICAL FEATURES:

→ middle Age, cyanosis, plethora, headache, dizziness, visual problem, thrombotic episodes.

LAB FINDINGS:

Peripheral smear:

→ Normocytic Normochromic

→ Hb ↑ed

→ RBC count ↑ed, WBC ↑ed, Platelet count ↑ed.

Bone Marrow:

→ Hypercellular → Hyperplasia → prominence of myeloid precursor. ↑ed Reticulin fibrosis of fibrosis.

Other findings:

→ Extramedullary hematopoiesis

→ ↑ Serum B12

→ JAK2 V617F mutation