POLYCYTHEMIA

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1- Relative polycythemia occurs in the setting of
A. Wilms’ tumor
B. Dehydration
C. Renal cell carcinoma
D. Polycythemia vera
E. Hypoxia
2- All of the following are causes of secondary absolute polycythemia, except:
A. Smoking
B. Renal artery stenosis
C. Polycystic kidney
D. Polycythemia vera
E. High altitude
3- The risk acute myeloid leukemia in polycythemia vera is
A. 2%
B. 10%
C. 30%
D. 50%
E. 70%
4- One of the following is a minor criterion for polycythemia vera
A. High hemoglobin
B. Hypercellular bone marrow
C. Low erythropoietin level
D. High erythropoietin level
E. Bone marrow fibrosis
Polycythemia denotes an abnormally high red cell count, usually with a corresponding increase in the hemoglobin level.
• Relative
• Absolute
  • Primary
  • Secondary
RELATIVE

- Decrease plasma volume with intact total RBC mass
  - Dehydration
  - Diarrhea
  - Diuretic therapy
ABSOLUTE

• Increase RBC mass
  • With high EPO (secondary)
  • Low EPO (primary)
- Hypoxia
  - Generalized
    - Smoking
    - High altitude
    - High affinity hemoglobins
  - Localized
    - Renal artery stenosis
    - Polycystic kidney disease

- Certain neoplasms
  - Wilms tumor
  - Renal cell carcinoma
  - Celebellar hemangioma
  - Hepatocellular carcinoma
• Low EPO

Polycythemia vera (PCV) is characterized by increased marrow production of red cells, granulocytes, and platelets (panmyelosis), but it is the increase in red cells (polycythemia) that is responsible for most of the clinical symptoms.
• Strongly associated with **JAK2 mutation**
• Valine-to-phenylalanine substitution at residue 617.
• Patients are prone to both thrombosis and bleeding.
• Splenomegaly mild at first severe in spent phase
• Hypercellular bone marrow.
• Late in the disease course, bone marrow fibrosis and significant organomegaly is present.
CLINICAL FEATURES

- Pruritis
- Headache dizziness
- Hyperuricemia and gout
- Increased risk of both major bleeding and thrombotic episodes.
  - Deep venous thrombosis
  - Stroke
  - Myocardial infarction
  - Bowel infarction
  - Budd-chiari syndrome
- Epistaxis and bleeding gums
- Major hemorrhage can occur in ~10% of the patients
• Phlebotomy and JAK2 inhibitors
• Spent phase; fibrosis and splenomegaly
• 2% might transform to acute myeloid leukemia
Criteria for diagnosis
(For diagnosis, all three major criteria or the first two major and the minor criterion)

**Major criteria**
1. Elevated hemoglobin (>16.5g/dl for men and >16 g/dL for women)
2. Bone marrow biopsy showing hypercellularity with increased trilineage growth (panmyelosis)
3. Presence of JAK2 V617F or other functionally similar mutation such as JAK2 exon 12 mutation

**Minor criteria**
1. Serum erythropoietin level below the reference range for normal