

Essential Thrombocythemia

Essential Thrombocythemia is caused by JAK2 mutations. It is often asymptomatic but can lead to thromboembolic events like spontaneous abortions, erythromelalgia, and gout. Diagnosis is made by isolated thrombocytosis on CBC and isolated megakaryocyte hyperplasia on bone marrow biopsy. Patients are often treated with prophylactic aspirin and hydroxyurea.



PLAY PICMONIC

Characteristics

JAK2 Mutation

[Jack in \(2\) Tutu Mutant](#)

Janus Kinase 2 is an intracellular tyrosine kinase involved in cytokine-mediated signaling. Mutations in the JAK2 gene are associated with myeloproliferative disorders like polycythemia vera, primary myelofibrosis, and essential thrombocythemia.

Presentation

Often Asymptomatic

[Thumbs-up](#)

Patients are usually asymptomatic for extended periods of time, and the disease may be discovered incidentally.

Thromboembolic Events

[Trombone with Thromboembolic Event](#)

Essential thrombocythemia can cause both micro- and macrovascular thrombosis. This can include transient ischemic attacks, deep vein thrombosis, erythromelalgia, and ocular migraine.

Spontaneous Abortion

[Spartan Aborting-fetus](#)

Abortion is an intrauterine death of the fetus before it attains viability. Due to the increased risk of thrombosis, there is a marked degree of uteroplacental insufficiency and thus, an increased risk of abortions.

Erythromelalgia

[Earth-red-molten-lava](#)

Erythromelalgia is characterized by a triad of redness, warmth, and burning pain of hands and feet. It is due to the decreased caliber of blood vessels causing vascular compromise to muscles and nerves.

Gout

[Gout-goat](#)

Due to the high cell turnover in essential thrombocythemia, serum uric acid is elevated which gets deposited in joints causing gouty arthritis.

Diagnosis

Thrombocytosis

Up-arrow Plate

Due to JAK2 mutation and increased megakaryocyte colony stimulating factors, platelet count is greater than 600,000/ μL in this disease.

Megakaryocyte Hyperplasia

Up-arrow Mega-carrot and Hiker-plates

Giant mature megakaryocytes with multilobulated nuclei are present in clusters in the bone marrow. This occurs from increased production and growth of platelet precursors (megakaryocytes) because of the mutation.

Management

Aspirin

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Aspirin is an irreversible cyclooxygenase-1 inhibitor and is an antiplatelet drug. It is given for the prophylaxis of thrombotic events in mild cases.

Hydroxyurea

Hydra-U-Rainbow

Hydroxyurea is an anti-cancer drug that arrests cells in the S phase of the cell cycle by inhibiting ribonucleotide reductase. It is given to reduce platelet counts.