The Blood Project

UNDERSTANDING ESSENTIAL THROMBOCYTHEMIA

A brief guide for patients with essential thrombocythemia (ET)

Essential thrombocythemia (ET) is a chronic blood cancer in which the bone marrow makes too many platelets. Platelets help the blood clot, but in ET they may become so numerous that they increase the risk of blood clots, and when extremely high they may also increase the risk of bleeding. Most people with ET feel well at diagnosis and remain stable for many years with treatment and regular follow-up. ET is a slow-moving myeloproliferative neoplasm, and with proper care most people can live long, active lives.

What are platelets?

Platelets are small blood cells that help stop bleeding when blood vessels are injured. They gather at sites of injury and form plugs that begin the clotting process. A normal platelet count is usually between 150,000 and 450,000. In ET, the bone marrow makes more platelets than the body needs.

What is essential thrombocythemia?

Essential thrombocythemia is a condition in which the bone marrow produces too many platelets because of changes in genes that regulate blood cell growth.

Most people with ET have one of three common gene changes called JAK2, CALR, or MPL mutations. These mutations are acquired during life and are not inherited in the usual sense. They are not contagious and are not caused by diet, stress, exercise, or lifestyle.

A smaller group of people have “triple-negative” ET, meaning none of the three common mutations are present. ET is typically long-term but very manageable with monitoring and treatment.

Why ET happens

ET happens because of acquired genetic changes inside bone marrow cells that disrupt the normal controls on platelet production. As a result, the marrow continues making platelets even when the body does not need them.

Important points include:

- acquired mutations such as JAK2, CALR, or MPL
- not usually inherited and not passed directly to family members
- not related to diet, stress, exercise, or lifestyle
- not contagious

Does it cause symptoms?

Many people with ET have no symptoms. Others may experience headaches, lightheadedness, or vision changes. Some notice numbness, tingling, redness, warmth, or burning pain in the hands or feet (erythromelalgia), which often improves with low-dose aspirin.

Fatigue is common. Some people develop fullness or discomfort in the left upper abdomen from an enlarged spleen.

When platelet counts become extremely high, platelets may stop working normally. This can cause nosebleeds, gum bleeding, or easy bruising. These bleeding symptoms are less common than clotting and are usually seen only at very high platelet levels.

Is it dangerous?

The main concern in ET is an increased risk of blood clots. Clots can occur in arteries or veins and may affect the brain, heart, lungs, legs, or abdominal veins.

Risk is higher in people who are age 60 or older, who have had a prior blood clot, or who have a JAK2 mutation. These factors help doctors determine who needs more intensive treatment.

When platelet counts rise to very high levels, often above about 1,000,000–1,500,000, bleeding risk can also increase. This happens because platelets may not function properly and certain clotting proteins can become depleted.

Over many years, a small percentage of people may develop myelofibrosis, and a smaller group may develop acute leukemia. Most people with ET never experience these complications, especially with modern treatment and regular follow-up.

How is it evaluated?

Evaluation usually includes:

- complete blood count to measure platelets and other blood cells
- genetic testing for JAK2, CALR, or MPL mutations
- review of personal and family history of clotting or bleeding
- physical examination, including checking spleen size
- iron studies, since iron deficiency can raise platelet counts
- bone marrow biopsy in selected situations to confirm the diagnosis or rule out related conditions

Do I need a bone marrow biopsy?

Not everyone with ET needs a bone marrow biopsy. Many diagnoses are made using blood counts and genetic testing alone.

A biopsy may be recommended if the diagnosis is uncertain, if no mutation is found (triple-negative ET), or if there is concern about another bone marrow condition. Your doctor will explain whether a biopsy is needed in your situation.

How is it treated?

Treatment focuses on reducing the risk of blood clots, managing symptoms, and keeping platelet counts in a safer range.

Low-dose aspirin is commonly used, especially for people with symptoms such as erythromelalgia or with cardiovascular risk factors. In very low-risk individuals, aspirin use may be individualized.

People at higher risk, or those with very high platelet counts or significant symptoms, may need medicines that lower platelet production. These include hydroxyurea, interferon, or anagrelide.

Hydroxyurea is most commonly used, while interferon is often preferred in younger patients or during pregnancy planning.

Managing blood pressure, cholesterol, diabetes, and avoiding smoking are also essential parts of reducing clot risk. Treatment plans are individualized and may change over time.

When should I contact my doctor?

Contact your doctor promptly if you develop new or worsening headaches, vision changes, chest pain, shortness of breath, numbness or tingling, leg pain or swelling, unusual bruising, nosebleeds, gum bleeding, black stools, or increasing fatigue.

Seek urgent medical care for sudden weakness, difficulty speaking, severe chest pain, coughing up blood, or uncontrolled bleeding. These may be signs of serious clotting or bleeding complications.

What is the usual plan going forward?

ET is a lifelong condition. Most people are followed with regular clinic visits and blood tests to monitor platelet counts, adjust medications, review symptoms, and reassess risk over time.
Many people remain stable for decades. Ongoing care focuses on clot prevention, symptom control, and early detection of any changes.

Key points to remember

- **chronic blood cancer:** ET is a long-term condition in which the bone marrow makes too many platelets.
- **acquired gene changes:** most cases are driven by acquired mutations such as JAK2, CALR, or MPL.
- **main risk is clots:** blood clots are the most important complication to prevent.
- **bleeding at very high counts:** extremely high platelet levels can also increase bleeding risk.
- **risk-based treatment:** age, clot history, and mutation status guide treatment decisions.
- **long-term outlook is good:** with regular follow-up and treatment, most people live long, active lives.