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Essential Thrombocythemia

Essential thrombocythemia (ET) is a chronic blood malignancy characterized by an abnormally high number of platelets in the blood. The increase in platelets can cause abnormal blood clotting in blood vessels and bleeding, and in late stages can progress to myelofibrosis (fibrous tissue build up in the bone marrow) or acute leukemia. ET is one of a group of rare bone marrow cancers called myeloproliferative neoplasms (MPNs) that develop due to an acquired gene mutation in the bone marrow stem cells.

Statistics

- JAK2 (V617F) (Janus kinase) gene mutations occur in about **50%** of patients with ET.
- Calreticulin (CALR) mutations occur in approximately **25% of patients** and MPL mutations occur in up to **five percent of patients. Ten percent are "triple negative,"** meaning they do not have any of these gene mutations.
- The incidence of ET in the United States is approximately **1.5 in every 100,000 people.**

Risk Factors

The exact causes of the acquired genetic mutation as seen in ET are not yet known. Risk factors may include:

- **Gender:** Women are one and a half times more likely to develop essential thrombocythemia than men.
- **Age:** A majority of patients diagnosed with ET are older than 60. Approximately one in five patients are under age 40.
- **Gene Mutations:** The JAK2 gene is most often mutated at V617F, followed in frequency by CALR and MPL mutations.
- **Blood Clotting:** Blood clotting is more common if JAK2 (V617F) is mutated.

Symptoms

Many patients with ET do not have symptoms when they are diagnosed. Diagnosis often occurs during a routine exam or blood test. However, some patients may experience persistent symptoms, including the following:

- Fatigue
- Migraines
- Visual or hearing disturbances
- Dizziness
- Coldness, blue discoloration, or pain in the fingers or toes
- Burning, redness, throbbing, and pain in the hands and feet
- Tingling
- Stroke
- Transient ischemic attack (TIA)
- Heart attack
- Deep vein thrombosis
- Pulmonary embolism (blood clot in lung)
- Easy bruising
- Numbness or weakness on one side of the body
- Abnormal bleeding such as nosebleeds, heavy menses, blood in urine, gastrointestinal bleeding
- Night sweats
- Discomfort in upper left side of abdomen caused by an enlarged spleen, occasionally with loss of appetite
- Unusual sites of blood clotting, such as veins inside the abdomen
- Weakness
- Fainting
- Weight loss
- Chest pain

Prevention

ET cannot be prevented. Research is underway to learn more about how the disease develops.

Treatment

ET patients who do not experience symptoms, are younger, and without cardiovascular comorbidities may not need active treatment other than monitoring. However, if symptoms are present or the patient has high risk features (age greater than 60, prior blood clots, or positive JAK 2 mutation), treatment may be needed. Treatment options include low-dose aspirin and other medications such as hydroxyurea, ruxolitinib, anagrelide, and interferon. Clinical trials can also be an important treatment option.

About Texas Oncology

With more than 530 physicians and 280 locations, Texas Oncology is an independent private practice that sees more than 71,000 new cancer patients each year. Founded in 1986, Texas Oncology provides comprehensive, multi-disciplinary care, and includes Texas Center for Proton Therapy, Texas Breast Specialists, Texas Colon & Rectal Specialists, Texas Oncology Surgical Specialists, Texas Urology Specialists, Texas Infusion and Imaging Center, and Texas Center for Interventional Surgery. Texas Oncology's robust community-based clinical trials and research program has contributed to the development of more than 100 FDA-approved cancer therapies. Learn more at www.TexasOncology.com.

Sources: Leukemia & Lymphoma Society, MPN Research Foundation, National Cancer Institute, National Center for Advancing Translational Sciences, and National Library of Medicine

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