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Essential Thrombocythemia

Essential thrombocythemia (ET) is a chronic blood malignancy characterized by an abnormally high number of platelets circulating in the blood. This increase in platelets can cause abnormal blood clotting in blood vessels as well as bleeding. 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. In late stages, ET can progress to myelofibrosis (fibrous tissue build up in the bone marrow) or, in rare cases, acute leukemia.

Statistics

- Janus kinase 2 (JAK2) V617F 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 ET than men.
- **Age:** A majority of patients diagnosed with ET are older than 60. Approximately one in five patients is 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
- Vision or hearing problems
- 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 (blood clot in the large veins in the legs)
- 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 do not have cardiovascular comorbidities may not need active treatment and may continue monitoring. However, if symptoms are present or the patient has high risk features (older than 60, prior blood clots, or JAK2 mutation) which may increase risk of blood clots, treatment may be needed. Treatment options include low-dose aspirin and other medications such as hydroxyurea, ruxolitinib, anagrelide, and interferon alfa. Other treatment options may be available through clinical trials.

About Texas Oncology

With more than 530 physicians and 280 locations, Texas Oncology is an independent private practice, a member of The US Oncology Network, that sees more than 71,000 new cancer patients each year. Founded in 1986, Texas Oncology provides comprehensive, multidisciplinary care, and includes Texas Center for Proton Therapy, Texas Breast Specialists, Texas Colon & Rectal Specialists, Texas Oncology Surgical Specialists, Texas Urology Specialists and Texas Infusion and Imaging Center. 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 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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