Cancer Association of South Africa (CANSA)

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Fact Sheet
on
Essential
Thrombocythaemia

Introduction

Essential thrombocythaemia (ET) is not a type of blood cancer as there are no cancerous cells. It is considered a chronic haematological malignancy and is sometimes referred to as a pre-leukaemic disorder which means that it may eventually cause one or other type of leukaemia. Essential thrombocythaemia (also known as essential thrombocytosis, essential thrombocythaemia, primarythrombocytosis) is a rare chronic blood disorder characterised by the overproduction of platelets by megakaryocytes in the bone marrow. It may, albeit rarely, develop into acute myeloid leukaemia or myelofibrosis.

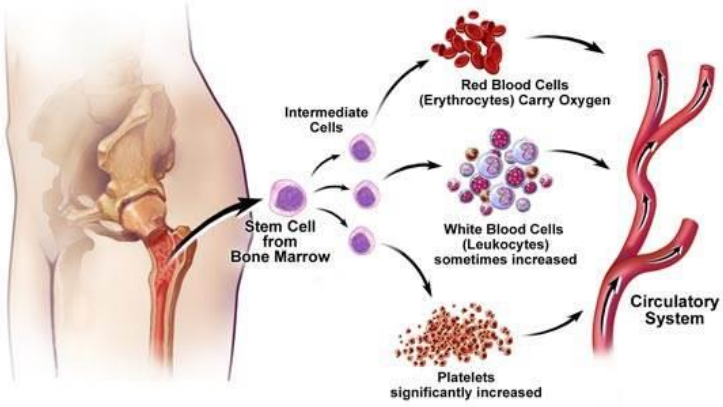
ET can result from a mutation in blood-forming stem cells, which causes blood cells (particularly platelets) to produce uncontrollably. However, research is still being done to understand the additional causes of ET. It is one of the known myeloproliferative neoplasms (MPNs).

Certain myeloproliferative neoplasms may become acute myeloid leukaemia (AML). Myeloproliferative neoplasms include chronic myelogenous leukaemia (CML), polycythaemia vera, primary myelofibrosis, essential thrombocythaemia, chronic neutrophilic leukaemia, and chronic eosinophilic leukaemia. Also called chronic myeloproliferative neoplasm.

The main types of MPN are:

- essential thrombocythaemia
- polycythaemia vera
- myelofibrosis.

[Picture Credit: Essential Thrombocythaemia]



ET is also known as essential thrombocytosis, thrombocytosis, and primary thrombocytosis.

Some people with essential thrombocythaemia develop myelo-fibrosis. This is a condition in which the bone marrow becomes scarred. People with MPNs rarely

develop leukaemia.
(Cancer Support Community; Myeloproliferative Disorder Research Consortium; Wikipedia).

To understand ET one must know about the role of bone marrow. Bone marrow is part of the immune system, which protects man from infection and disease. The marrow is in the spongy material found inside bones. This is where stem cells are made. Stem cells are blood cells at their earliest stage of development. All blood cells grow from stem cells. The three main types of blood cells are:

- red blood cells – which carry oxygen around the body
- white blood cells – which fight infections
- platelets – which help blood to clot to prevent bleeding.

The bone marrow makes millions of blood cells every day. When the cells are mature they are released into the blood stream. In MPNs, the bone marrow makes too many of one or more types of blood cells (Macmillan Cancer Support).

Incidence of Essential Thrombocythaemia in South Africa
The National Cancer Registry (2012) does not provide any information regarding the incidence of Essential Thrombocythaemia in South Africa.

Affected Populations
Fewer than 1 in 100 000 individuals are diagnosed with ET. Women are more likely to be diagnosed with ET than men, although the reason for this is unknown.

The average age of onset is mid-fifties, but the range is wide, and includes women in their childbearing years, which makes up an important subset of ET patients with special therapeutic considerations. In children ET is exceedingly rare and typically is an inherited genetic disorder. In adults, the genetic mutations typically identified in ET are not inherited, and instead are acquired genetic accidents (known as an acquired mutation) that happen during an individual's lifetime. Genetic accidents happen to all as part of ageing, although it does not always result in a disease.
(National Organization for Rare Diseases).

Causes of Essential Thrombocythaemia (ET)
The causes of ET are still unknown. About half of ET patients have a mutation called JAK2V617F within the blood-forming cells, which leads to many characteristic features of the disease.

Some risk factors associated with ET include:

- Gender - women are 1.5 times more likely than men to develop the condition
- Age - people older than 60 are most likely to develop the condition, although 20 percent of those affected are younger than 40
- Environment - exposure to chemicals or to electrical wiring may increase an individual's risk for the condition

In all of the myeloproliferative disorders, a stem cell that is capable of producing red blood cells, certain white cells and platelets somehow goes haywire and no longer keeps the blood elements produced by the marrow in balance. In essential thrombocythaemia, the marrow produces too many platelets.
(Johns Hopkins Medicine; Myeloproliferative Disorder Research Consortium).

Symptoms of Essential Thrombocythaemia (ET)
Many patients do not have any symptoms. The diagnosis is often made after blood counts were done as part of a routine check-up which then reveals a high platelet count. Symptoms, if present, include fatigue, or blood vessel disturbances or bleeding.

Blood vessel disturbances or bleeding can result in:

- Headache
- Vision disturbances or silent migraines
- Dizziness or lightheadedness
- Fainting
- Coldness or blueness of fingers or toes
- Burning, redness, and pain or numbness in the hands and feet
- Redness, throbbing and burning pain in the hands and feet (erythromelalgia)
- Mildly enlarged spleen
- Difficulty speaking or understanding speech (aphasia)
- Blurred, double or decreased vision
- Easy bruising, nosebleeds or heavy periods
- Gastrointestinal bleeding or blood in the urine

Erythromelalgia is a rare and frequently devastating disorder that typically affects the skin of the feet or hands, or both, and causes visible redness, intense heat and burning pain. It usually affects the lower extremities (legs and feet) and upper extremities (arms and hands) including other body parts like the face, ear or nose. It usually affects both sides of the body, but has been known to only affect one side. The association of pain and burning sensations can be extremely severe.
(Johns Hopkins Medicine; Mayo Clinic; Erythromelalgia Association).

There are also other metabolic abnormalities found in patients with myeloproliferative neoplasms. These include:

- Elevated uric acid counts are seen in about half of MPN patients during the course of their disease. If untreated, this leads to uric acid stones, uric acid neuropathy, acute gout, and chronic gouty arthritis. Patients may experience joint pain as a result
- Low cholesterol levels (hypocholestoraemia), particularly in those with enlarged spleens (splenomegaly)
- Elevated histamine levels. Symptoms of increased histamine release include pruritis (itching characteristically produced by bathing or showering), heartburn, acid eructation (gas) , peptic ulcer, small bowel hypermotility, flushing and angioneurotic oedema (swelling of skin, mucous membranes or viscera). This occurs in 2/3rds of myeloproliferative disorder patients and correlates with presence of elevated basophil count and hyperhistaminaemia

- Hypermetabolism which commonly manifests as weakness and fatigue in the absence of anaemia

(Myeloproliferative Disorder Research Consortium).

Complications of Essential Thrombocythaemia (ET)

The abnormal blood clotting of essential thrombocythaemia can lead to a variety of potentially serious complications, including:

- Pregnancy complications. While many women who have thrombocythaemia have normal, healthy pregnancies, they must ensure that their doctor regularly monitors their condition. Uncontrolled thrombocythaemia may cause miscarriage, premature delivery, high blood pressure (preeclampsia), early separation of the placenta from the uterine wall (placental abruption) and slow foetal growth. The risk of complications may be reduced with regular check-ups and medication
- Stroke. A clot that blocks blood flow to the brain can cause a stroke. If someone develops signs and symptoms of a stroke, get immediate medical attention
- Heart attack. A clot that obstructs blood flow to the heart can cause a heart attack. If someone develop signs and symptoms of a heart attack, such as pressure, fullness or a squeezing pain in the centre of the chest lasting more than a few minutes; pain extending to the shoulder, arm, back, teeth or jaw; shortness of breath; and sweating or clammy skin, get immediate medical attention
- Essential thrombocythaemia can also cause bleeding (haemorrhage) with significant blood loss. A small minority of people with essential thrombocythaemia may later develop acute leukaemia or myelofibrosis, both of which can be life-threatening:
    - Acute leukaemia. Acute myelogenous leukaemia is a type of blood and bone marrow cancer that progresses rapidly
    - Myelofibrosis. This progressive bone marrow disorder results in bone marrow scarring, severe anaemia, and enlargement of your liver and spleen

(Mayo Clinic).

Treatment of Essential Thrombocythaemia (ET)

ET is treated based on the person's risk of complications, such as bleeding and blood clotting. People with no signs or symptoms other than an increased platelet count have a lower risk of complications. These people, especially those with no other cardiovascular risk factors, may only need regular check-ups.

People at high risk for bleeding and clotting complications need treatment.

Risks for clotting complications include:

- previous clot

- cardiovascular risk factors – high cholesterol, diabetes, smoking, obesity or high blood pressure

Risks for bleeding complications include:

- extremely high platelet count
- regular use of standard-dose aspirin or non-steroidal anti-inflammatory drugs

(Canadian Cancer Society).

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Sources and References

Canadian Cancer Society
http://www.cancer.ca/en/cancer-information/cancer-type/leukemia/pathology-and-staging/essential-thrombocythemia/?region=on

Cancer Support Community
http://www.cancersupportcommunity.org/MainMenu/About-Cancer/Types-of-Cancer/Myeloproliferative-Neoplasms-2/What-is-Essential-thrombocythemia-ET.html

Erythromelalgia Assocation
http://www.erythromelalgia.org/WhatisEM.aspx

Essential Thrombocythaemia
https://www.google.co.za/search?q=essential+thrombocythemia+cancer&source=lnms&tbm=isch&sa=X&ei=soSyU5oR8ujsBoqAgfgG&sqi=2&ved=0CAYQ_AUoAQ&biw=1517&bih=714&dpr=0.9#facrc=_&imgdii=_&imgrc=J7h5nTaOV-c-TM%253A%3BxT2sEFx81lriCM%3Bhttp%253A%252F%252Fstatic.cdn-seekingalpha.com%252Fuploads%252F2014%252F1%252F834108_13906209951263_rId8.jpg%3Bhttp%253A%252F%252Fseekingalpha.com%252Farticle%252F1971101-the-acquisition-of-mills-pharmaceuticals-put-another-arrow-in-the-quiver-of-galena%3B544%3B319

Johns Hopkins Medicine
http://www.hopkinsmedicine.org/kimmel_cancer_center/centers/bone_marrow_failure_disorders/essential_thrombocytosis.html

MacMillan Cancer Support
http://www.macmillan.org.uk/Cancerinformation/Causesriskfactors/Pre-cancerous/Essentialthrombocythaemia.aspx

Mayo Clinic
http://www.mayoclinic.org/diseases-conditions/thrombocythemia/basics/symptoms/con-20034386
http://www.mayoclinic.org/diseases-conditions/thrombocythemia/basics/complications/con-20034386

Myeloproliferative Disorder Research Consortium
http://www.mpd-rc.org/faq.php?cat_id=4

National Organization for Rare Diseases
https://rarediseases.org/rare-diseases/essential-thrombocythemia/

Wikipedia
https://en.wikipedia.org/wiki/Essential_thrombocytosis