

Essential Thrombocythemia

Your doctor has just told you that you have an essential thrombocythemia.

Thrombocythemia is the word used for a collection of blood diseases which affect mainly your blood platelets. These cells are produced by the bone marrow and are essential for coagulation.

In 85 % to 90 % of cases, Thrombocythemias are called "reactive" as they occur especially after intensive stress, an infection, an inflammation, a solid tumour, an iron deficiency...

In 10 % to 15 % of cases, they are called "essential" as they are not the consequence of another problem.

Essential Thrombocythemia is one of the blood diseases grouped under the name of "myeloproliferative syndromes". It is characterised by persistent excessive production of blood platelets within the bone marrow. Platelets play an important role in coagulation of the blood and in the phenomena of haemostasis (the stopping of bleeding in a vascular wound).

The way in which this disease occurs is not very well understood.

A high number of platelets can mainly lead to two kinds of complications:

- ♦ The occurrence of a thrombosis, i.e. the formation of a bloodclot that partially or totally obstructs a blood vessel (artery or vein) Depending on the part of the body or the organ concerned, a thrombosis can have major or minor consequences. In the case of the heart, it can induce a myocardial infarction; in the brain, it can cause what is called a TIA (Transitory Ischaemic Attack), causing effects such as migraine, visual problems (double vision, changes in the field of vision etc) or even a stroke. A thrombosis may also induce phlebitis - inflammation of a vein at the place where the clot is.

However, most often a thrombosis associated with essential thrombocythemia affects the microcirculation, i.e. the very little veins, producing a syndrome called erythromelalgia. This results in the appearance in hands and feet of redness and painful burning sensations.

- ♦ The occurrence of a haemorrhage, i.e. bleeding of the skin or mucous membrane. In most instances, this will show itself in nose bleeds or bleeding of the gums, bruising or the presence of blood in the urine. Internal haemorrhaging can occur (especially in the digestive system) but this is much more rare. Essential thrombocythemia is a fairly rare disease, as there are 1 to 2.5 cases per 100,000 population counted each year in France. It occurs most often after the age of 50, with men and women affected equally. However, one notices a peak in the frequency around the age of 30, especially among women.

The first symptoms

Essential thrombocythemia may not show any clinical symptoms for a long time. So when it is diagnosed in half the cases, it is asymptomatic.

In the other cases, essential thrombocythemia can reveal itself through different symptoms, more or less severe, like a thrombosis causing phlebitis in a leg, a cardiac or neuralgic problem or bleeding nose or gum, bruising occurring after a slight knock, or even for no apparent reason. Erythromelalgia - reddening and burning sensations in extremities- is one of the key symptoms indicating the disease. In every case it is seeing the doctor that leads to the search for the cause of these various symptoms.

Diagnosis

When it is symptom-free, essential thrombocythemia is generally discovered by pure chance, in the course of a blood test. The analysis shows the high level of platelets in the blood, generally greater than $600000/\text{mm}^3$ (whereas the norm is between $150\ 000$ and $400\ 000/\text{mm}^3$), and possibly reaching several million per mm^3 . Most often, there is no other blood abnormality; levels of haemoglobin, red cells and white cells remain within the normal ranges.

The diagnosis is based in the first place on a complete clinical examination which aims mainly to look for an increase in the volume of the spleen (splenomegaly) that is present in half the number of cases, and in that of the liver (hepatomegaly) present in 15 to 20% of patients. At the same time, different blood tests are carried out looking especially for an iron deficiency. Finally a puncture, or even a bone marrow biopsy (removal under local anaesthetic of a sample of bone marrow), may be carried out. Studying the sample under a microscope enables the cells at the origin of the blood cells to be analysed and any abnormalities present to be detected. It also anything unusual about the chromosomes in the marrow cells to be looked into. All these tests are aimed above all at seeing if the increase in the number of platelets is not linked to some other disease, as there is specific test for the diagnosis of essential thrombocythemia. Thus a genetic mutation of the cells in the bone marrow (mutation of the JAK2 gene) is found in only half the cases.

Certain treatments may lead to sterility, especially where men are concerned. So sperm conservation is offered after the diagnosis.

Any treatment is likely to produce unwanted side-effects and may present risks. Your doctor will inform you and will tell you which signs to look out for before you start the treatment suggested

Participating in a clinical trial

The best way to contribute to the improvement of disease management is to treat patients in the context of clinical trial. If your doctor suggests this could apply to you, he will explain its purpose, protocol, expected benefits, potential risks and will give you an information leaflet.

Participating in a trial of course means you will first have to give your written informed consent.

Treatment

Treatment of essential thrombocythemia targets on one hand prevention of the occurrence of a thrombosis or haemorrhage and on the other to decrease the number of platelets in the blood if necessary.

Prevention of thromboses is achieved in essence by taking aspirin, as it has anticoagulant properties, i.e. limiting the formation of blood clots. However, as this medicine can also prompt haemorrhages, it is prescribed at a low dosage.

The reduction in the number of platelets can be achieved thanks to medicines called "cytoreductive agents". At present several medicines of this kind are available. The choice of one rather than another is made in the light of the level of their tolerance expected from each individual patient.

The methods chosen for treatment depend on several factors. To begin with, the stage the disease has reached (in terms of the number of platelets and whether a thrombosis has occurred previously), the age of the patient and the cardiovascular risks that he presents (obesity, smoking, diabetes, high levels of fat in the blood). In the case of young patients showing no symptoms and with no cardiovascular risks, simply monitoring, and a low dosage of aspirin is generally suggested. For older patients and those who present a significant risk of thrombosis, treatment with a cytoreductive agent linked with low dose aspirin is recommended.

When treatment is initiated, currently available medicines enable the disease to be controlled in the great majority of cases, with side effects that are more or less pronounced according to the product used and the sensitivity of each individual patient.

The life expectation of patients affected by essential thrombocythemia is therefore similar to that of people who are well.

Contraception and Pregnancy

Essential thrombocythemia may affect young women who wish to have available a method of contraception or on the other hand may wish to have a child. For contraception, it is recommended to use a pill without oestrogen, containing only a progestative agent that is non-thrombosis inducing. Use of an inter-uterine device is not compatible with the prescription for aspirin, as that increases the risk of haemorrhage, and is therefore not recommended.

Having a child is completely conceivable for these patients, but it requires specialised medical monitoring and adjustment to the treatment. Moreover, essential thrombocythemia is associated with an increased risk of miscarriage, especially in the first three months of pregnancy.

Useful contacts:

- **Secretarial / appointment:**
- **Nursing consultation:**
- **Consulting psychologist:**
- **Social worker:**
- **In an emergency:**