

Bone marrow aplasia

You have just been diagnosed with aplastic anaemia also known as bone marrow aplasia.

The bone marrow is the soft tissue inside your bones which is responsible for the production of all types of blood cells.

'Aplasia' means the production of blood cells in the bone marrow has sharply decreased, so that your blood count is low.

The bone marrow produces such blood cells as red blood cells, white blood cells and platelets. Red blood cells carry oxygen in the blood and circulate it around the body while white blood cells fight infections and platelets are essential to ensure blood clotting and help stop bleeding. These three types of cells are produced within the bone marrow by so-called 'hematopoietic cells'.

In aplastic anaemia the bone marrow hematopoietic cell count is gradually lowered resulting in the production of fewer blood cells, with of course variations from one person to another, but still inducing a sharp drop of the blood count.

In most cases the origin of aplastic anaemia is unknown; the disease is then said to be idiopathic. It is commonly assumed to be due to some auto-immune event; for some unknown reason the body's immune system destroys the body's own blood cells.

This disease strikes both men and women of all ages although two age-groups have an increased risk : both young adults (around 20-25 years of age) and older people (over the age of 50) have higher rates of aplastic anaemia. It is, however, a rare disease with only 120 new cases a year in France.

Aplastic anaemia is not contagious, and only some rare forms like Fanconi syndrome are hereditary and diagnosed in childhood.

Medecin's notes :

Clinical features

Aplastic anaemia may occur suddenly or very gradually. The lower blood count is responsible for three main signs :

- Anaemia, i.e. a low red blood cell count. Its symptoms include pallor, fatigue, breathlessness and palpitations on exertion.
- Repeated infections. The low white blood cell count weakens the immune system so that the body is less apt to fend off infections, however slight, which therefore become more frequent and sometimes more severe.
- Bleeding. When the platelets become less numerous (thrombopenia), bleedings of varying severity may occur, affecting mostly the nose and the gums. Ecchymoses and hematomas (commonly called bruises) may occur for some unexplained reason.

Diagnosis

The diagnosis of aplastic anaemia first relies on a full blood cell count made from a simple blood test in order to count the different blood cells. The result shows that their numbers are much lower than the normal range.

To confirm the diagnosis a bone marrow aspirate (a myelogramme) has to be performed. It is performed under local anaesthesia and consists in inserting a hollow needle into a bone, usually either the breastbone or the hipbone. A small quantity of bone marrow is then removed and examined under a microscope. If aplastic anaemia is present, this examination will reveal very few or no hematopoietic cells.

Definitive confirmation of the diagnosis requires an osteomedullary biopsy which consists in sampling both bone tissue and bone marrow under local anaesthesia. The microscopic study of bone tissue allows to realise how depleted the bone marrow is and to rule out other diseases.

Bone marrow aplasia (suite)

Daily advice

People with aplastic anaemia are advised to watch their everyday life in order to avoid the risks of complications due to their low blood cell count. Here is some advice :

- do not eat foods of unknown origin
- wash your hands regularly, especially before meals
- do not visit anyone with a contagious disease
- avoid situations where you may get knocks and blows (such as some sports) to prevent bleeding

Moreover, should fever appear, immediately get in touch with your haematologist or your GP so that they can help you fight against some possible infection.

Your treatment may induce adverse effects and carry risks. Your doctor will keep you informed and tell you what symptoms to watch out for before you start on the suggested treatment.

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.

Useful contacts:

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

Management

The disease may progress differently from one person to another. When the symptoms are moderate, simple medical supervision may be enough. But most often repeated infections and bleedings require treatment.

To date two treatments are used:

- Immunosuppressive treatment: Immunosuppressives are drugs which inhibit the patient's immune system, thus reducing, and even stopping, the destruction of stem cells. In most cases the treatment combines two immunosuppressives: anti-leukocyte serum (ALS) and a selective immunosuppressive drug. The former is administered through an intravenous drip over five days while the latter is taken orally twice a day over several months. Corticosteroids are usually taken at the same time as SAL infusions to improve tolerance. Patients are usually hospitalised at the beginning of the treatment to protect them from infections due to immunosuppression. The course of the treatment lasts at least three months, but can be extended for over a year depending on disease progress. This treatment does not apply to the rare congenital forms of the disease.

This immunosuppressive treatment is efficient in most patients, however not in all of them. In case of a good response the disease and its symptoms improve or are stalled. However, relapse is still possible several years later.

- Hematopoietic stem cell allograft may lead to healing. The graft consists in replacing the patient's immune cells and remaining stem cells with a donor's stem and immune cells. These new disease-free cells allow to restore a normal blood count. This type of graft requires finding a matching donor (within the patient's family or on a donor list).

This treatment is heavy and may lead to serious complications; it requires a hospital stay in a sterile room for several weeks, which is why it is offered only to young patients (under 45/50 years-old) with a severe form of the disease.

Meanwhile other treatments may be required to fight the symptoms of the disease, such as antibiotics to prevent or treat infections. In case of marked anaemia or thrombopenia transfusions with red blood cells or platelets are often required.

Follow-up

Regular visits with your haematologist and your general practitioner are necessary throughout the treatment course, but their frequency may vary according to each patient's evolution.