

Severe Aplastic Anaemia Patient Information

What is Severe Aplastic Anaemia?

Severe Aplastic Anaemia is a rare, non contagious and potentially life threatening disorder that results from unexplained failure of the bone marrow to produce red blood cells, white blood cells and platelets. A decrease in the production of these blood cells means the patient is more at risk of bleeding, tiredness and infections.

How often does it occur?

Severe Aplastic Anaemia affects 2- 6 people per million worldwide. It is more common in Asia than in the western world and most cases occur in adults.

What causes Severe Aplastic Anaemia?

The cause of the disease is usually unknown and is often termed "idiopathic". It is thought that most cases of Severe Aplastic Anaemia are "acquired", that is it is not inherited, it is not present from birth and develops some time during life.

Idiopathic Acquired Severe Aplastic Anaemia is thought to be caused by an immune reaction where the body's immune cells become confused and start to attack body tissue with no clear underlying cause. In particular the bone marrow cells are eradicated by the immune attack. The bone marrow makes blood.

In a few patients exposure to toxins / certain drugs / certain disease or infections may trigger the development of Severe Aplastic Anaemia. Whenever a patient is given drugs known to carry a risk of causing Severe Aplastic Anaemia, the patient is followed very closely and will have regular blood tests. Some drugs which are thought to lead to rare cases of Severe Aplastic Anaemia are: anti-cancer drugs, immunosuppressants, drugs used to treat rheumatoid arthritis and some antibiotics.

What are the signs and symptoms?

The signs and symptoms seen most often include: tiredness, paleness, shortness of breath on exertion, rapid heart beat, infections, rash, easy bruising, bleeding gums, and prolonged bleeding.

Tiredness and weakness are caused by anaemia (low red cell numbers); bruising and or bleeding problems result from a low platelet count and infections are a result from low numbers of healthy white cells.

What tests are done?

Blood tests are done to measure the different types of blood cells and the haemoglobin level. (Refer to blood section for a full description of the function of each blood cell.)

Bone Marrow Biopsy: this involves taking a small amount of marrow from inside the bone. This procedure is done under a General Anaesthetic. (See Bone Marrow Biopsy section for a full description of the procedure.)

The main finding which defines Severe Aplastic Anaemia is that the bone marrow is lacking most or all of the normal cells. The few blood producing cells which are present appear normal.

Chromosome analysis this may be done on the cells of the blood or the bone marrow. Other diseases of the bone marrow show changes in the chromosome of marrow cells and in Severe Aplastic Anaemia these changes are not seen except in extremely rare cases.

Classification of Severe Aplastic Anaemia

Aplastic Anaemia can be classified as mild or severe based on the test results. The condition is classed as severe if two out of three of the following are present:

- Absolute neutrophil count less than $500 \times 10^9/L$
- Platelet count less than $20 \times 10^9/L$
- Reticulocytes (immature red cells) less than $25 \times 10^9/L$
- Bone marrow has markedly reduced numbers of blood producing cells.

Severe Aplastic Anaemia is a life threatening condition. Studies have shown that mortality one year after diagnosis is more than 80% if not treated. Less severe disease has a better prognosis. As factors change for each individual it is important that you speak to your doctor about your child's specific information.

How is Aplastic Anaemia Treated?

Types of treatment can include:

- Supportive therapy - transfusions, treatment of infection
- Immunosuppressive therapy
- Bone marrow transplantation

- **Supportive therapy**

Patients with Severe Aplastic Anaemia require transfusion support until the diagnosis is made and specific treatment can be started. Both red cell and platelet transfusions are often required. Infections are the major risk which may lead to death in patients with Severe Aplastic Anaemia. Patients are at risk of infections due to their prolonged very low white cell count. All infections require prompt therapy with specific antibiotics. Blood forming growth factor drugs, such as G-CSF, may be used to promote white cell recovery.

- **Immunosuppressive therapy**

Drugs which suppress the immune system may be used in patients with Severe Aplastic Anaemia. Special antibodies called ATG (anti-thymocyte globin) are used. These antibodies reduce the activity of the lymphocytes which are attacking the bone marrow cells. A drug called cyclosporin may also be used and this drug affects the T-lymphocytes specifically. Other drugs that can be used include corticosteroids and blood forming growth factors such as G-CSF. The response to immunosuppressive therapy can be slow.

- **Bone Marrow Transplant**

Bone marrow transplant is the other main treatment option for patients with Severe Aplastic Anaemia. There is a significant risk of death and illness with bone marrow transplant. The overall risk from bone marrow transplant is similar to the overall risk from immunosuppressive therapy.

Family members are tested for their "HLA (Human Leucocyte Antigen)" type to see if they match the patient. A fully matched family member could be a potential bone marrow donor. If no match is found an unrelated donor from the bone marrow registry may be sought.

Bone marrow transplant involves using drugs and /or radiotherapy to “condition” the bone marrow (remove all cells and disease) to allow the donated stem cells or bone marrow to grow. If bone marrow transplant is the treatment for your child you will be given specific education and information about the whole process from the health care team.

Insertion of a Central Venous Access Device.

As the treatment and support therapy is intense it is essential that venous access is available. Your child will have a central venous catheter inserted to enable direct venous access to give required treatment. Central venous catheters are explained fully in the Central Venous Access Device section.

References:

Aplastic Anaemia Facts And Statistics – E Medicine
Aplastic Anaemia – Leukaemia Research Fund Booklet
Aplastic Anaemia Disease Information – St Jude Children’s Research Hospital
National Organisation for Rare Disorders, Inc – Aplastic Anaemia