

WHAT IS APLASTIC ANEMIA?

Aplastic anemia is a rare but serious disease of the bone marrow. Each year, between 600 and 900 people in the United States are diagnosed with it. Read below for more information about the cause, treatment, and prognosis of aplastic anemia.

This "Patient Education" tear sheet was produced in collaboration with the Aplastic Anemia & MDS International Foundation (AA&MDSIF) (www.aamds.org).

What is Aplastic Anemia?

Aplastic anemia is a disease of the bone marrow that occurs when the bone marrow stops producing enough new blood cells. Bone marrow is a sponge-like tissue inside the bones that makes stem cells that develop into red blood cells, white blood cells, and platelets.

In people who have aplastic anemia, the bone marrow's stem cells are damaged. Any blood cells that the marrow does make are normal, but there are not enough of them.

What Causes Aplastic Anemia?

Aplastic anemia is caused by destruction of the blood-forming stem cells in a person's bone marrow. Most research suggests that stem cell destruction occurs because the body's immune system attacks its own cells by mistake. Normally, the immune system attacks only foreign substances. When the immune system attacks one's own body, it is known as an autoimmune disease; aplastic anemia is generally thought to be an autoimmune disease.

Aplastic anemia can be either *acquired* or *inherited*. "Acquired" means a person is not born with the condition, but he or she develops it. "Inherited" means a person's parents passed the gene for the condition on to the individual.

Acquired aplastic anemia can begin any time in life. About 75 out of 100 cases of acquired aplastic anemia are idiopathic, meaning they have no known cause.

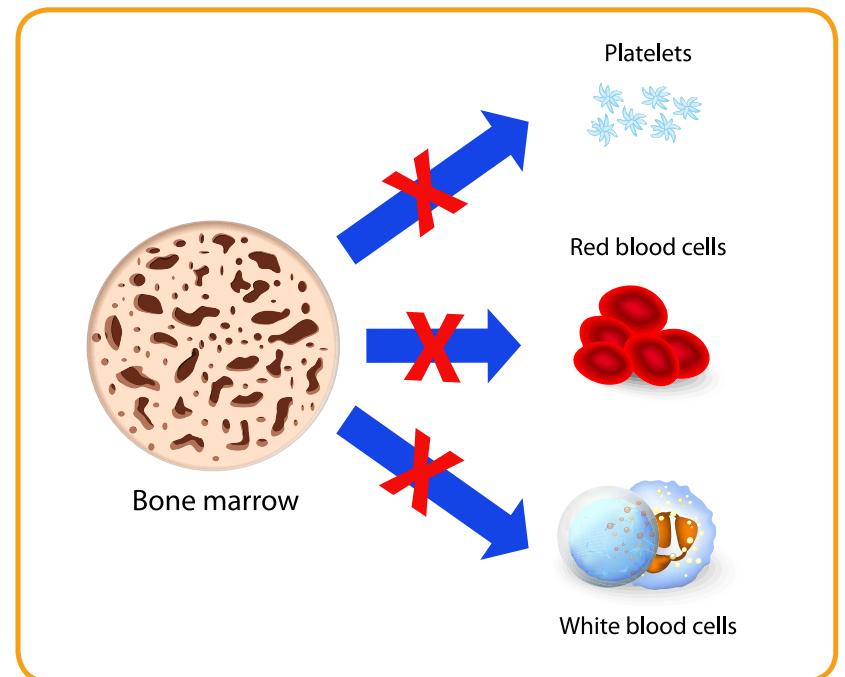
Inherited aplastic anemia is passed down through the genes from parent to child. It is usually diagnosed in childhood and is much less common than acquired aplastic anemia. People who develop hereditary aplastic anemia may have other genetic or developmental abnormalities. For instance, certain inherited conditions can damage the stem cells and lead to aplastic anemia. Examples include Fanconi anemia, Shwachman-Diamond syndrome, dyskeratosis congenita, and Diamond-Blackfan anemia.

About 25 out of 100 cases of acquired aplastic anemia can be linked to one of several causes. These include:

- ▶ Toxins, such as pesticides, arsenic, and benzene
- ▶ Radiation and chemotherapy used to treat cancer
- ▶ Treatments for other autoimmune diseases, such as lupus and rheumatoid arthritis
- ▶ Pregnancy (sometimes, this type of aplastic anemia improves on its own after the woman gives birth)
- ▶ Infectious diseases, such as hepatitis, Epstein-Barr virus, cytomegalovirus, parvovirus B19, and HIV
- ▶ Sometimes, cancer from another part of the body can spread to the bone and cause aplastic anemia.

What Are the Symptoms of Aplastic Anemia?

The symptoms of aplastic anemia are caused by low blood cell counts. The symptoms depend on which type of blood cell is affected.



Low red blood cell count: The most common symptom of a low red blood cell count is fatigue. A low red blood cell count also can cause shortness of breath; dizziness, especially when standing up; headaches; coldness in your hands or feet; pale skin; and chest pain.

Low white blood cell count: Also called neutropenia, a low white blood cell count can increase the risk for infections.

Low platelet count: A low platelet count, also called thrombocytopenia, can lead to bleeding problems and cause a person to bruise easily.

How Is Aplastic Anemia Diagnosed?

Diagnosing aplastic anemia can be a complex process. Doctors use three main tools to help them diagnose and understand each case of aplastic anemia. These include blood tests, medical history, and bone marrow examination.

Blood Tests

When trying to figure out the cause of a person's symptoms, the doctor will ask for blood samples. These samples will be used in a number of tests, including:

- A complete blood count (CBC) test to measure the number of each blood cell type in a person's blood sample
- A reticulocyte count to measure the number of young red blood cells in a person's blood
- An EPO, or erythropoietin, count. EPO is a protein made in a person's kidneys that causes the bone marrow to make more red blood cells
- Iron level measurements
- Vitamin B-12 and folate levels. A shortage of these vitamins reduces blood cell production in the bone marrow and causes a drop in the number of white blood cells, red blood cells, and platelets.

Medical History

To understand what is causing a person's symptoms and low blood counts, the doctor will take a detailed medical history. He or she may ask questions such as:

Where Can I Get More Information?

The Aplastic Anemia & MDS International Foundation (AA&MDSIF) is a resource for aplastic anemia patients and their caregivers. Below are a few different ways to get more information and assistance:

• **Toll-free phone number:** (800) 747-2820

• **Email:** info@aamds.org

• **Website:** www.AAMDS.org

AA&MDSIF OFFERS THE FOLLOWING SERVICES:

- Free support from Patient Education Specialists
- Free educational materials on many topics related to MDS
- Online Learning Center with programs on all aspects of MDS
- Conferences for patients and family members
- Clinical trials information
- Newsletters and E-News Alerts with important information and updates
- One-to-One Support Connection for patients interested in speaking with other MDS patients

- What are your symptoms?
- What medications or herbal supplements have you been taking?
- Have you been exposed to harmful chemicals?
- Did you have chemotherapy or radiation treatments in the past?

Bone Marrow Examination

A person's primary doctor or another doctor called a pathologist will take a bone marrow sample from the hip bone to examine. The doctor will look at the liquid bone marrow under a microscope and send a sample of the bone marrow to a lab. The bone marrow test shows:

- The quantity (cellularity) of a person's bone marrow occupied by different cells
- Exactly what types and amounts of cells a person's bone marrow is making
- Increased, decreased, or normal levels of iron in a person's bone marrow
- Chromosomal (DNA) abnormalities

Doctors classify aplastic anemia into three groups, based largely on the patient's neutrophil count. Neutrophils are a type of white blood cell that fight bacterial infections. These types include:

- **Moderate aplastic anemia:** Blood counts are low, but not as low as with severe aplastic anemia. A person may have few or no symptoms.
- **Severe aplastic anemia:** Neutro-

phil count is less than 500 cells per microliter.

- **Very severe aplastic anemia:** Neutrophil count is less than 200 cells per microliter.

How is Aplastic Anemia Treated?

The main goal of aplastic anemia treatment is to increase the number of healthy cells in the blood. When blood counts go up:

- A person is less likely to need blood from a donor (transfusion)
- Quality of life gets better
- Symptoms are not as bad

People who have mild or moderate aplastic anemia may not need treatment as long as the condition doesn't get worse. People who have severe aplastic anemia need medical treatment right away to prevent complications.

People who have very severe aplastic anemia need emergency medical care in a hospital. Very severe aplastic anemia can be fatal if not treated right away.

Removing a known cause of aplastic anemia, such as exposure to a toxin, also may cure the condition.

Treatment for aplastic anemia falls into three categories:

- **Supportive care,** or treatments that help to manage the symptoms of aplastic anemia and is not a cure. This approach includes the use of blood transfusions, iron chelation therapy

to treat iron overload, growth factors, and antibiotics.

- **Immunosuppressive drug therapy** lowers the body's immune response to prevent one's immune system from attacking the bone marrow, let stem cells grow back, and raise blood counts. In acquired aplastic anemia, immunosuppressive therapy with anti-thymocyte globulin (ATG) plus cyclosporine is the therapy of choice for older patients and patients who do not have a matched stem cell donor. For patients with severe aplastic anemia who are not candidates for stem cell transplant eltrombopag is approved for use. Eltrombopag works by helping to increase production of blood cells.

- **Stem cell transplantation** replaces damaged stem cells with healthy ones from another person (a donor). During the transplant, which is similar to a blood transfusion, a person gets donated stem cells through a tube placed in a vein in the chest. The stem cells travel to the bone marrow and begin making new blood cells. Stem cell transplants may cure aplastic anemia in people who are eligible for this type of treatment. The transplant works best in children and young adults with severe aplastic anemia who are in good health and who have matched donors, while older people may be less able to handle the treatments needed to prepare the body for the transplant.

Special Considerations for Patients with Aplastic Anemia

Everyday events can be more risky for those diagnosed with aplastic anemia than for healthy people. Here are some examples.

- ▶ **Airplane travel and high altitudes:** Flying in an airplane or going up high in the mountains may cause a shortage of oxygen or chest pain. Before doing either of these things, it's a good idea to get a red blood cell count and a platelet count, get necessary transfusions, and ask the doctor if it is safe.
- ▶ **Dental care:** Brush and floss teeth and get regular dental care to reduce the risk of infections in the mouth and throat. If a dental procedure is needed, talk with the treating physician first as antibiotic or other treatments may be needed.
- ▶ **Physical activity:** Talk with the doctor about what types and amounts of physical activity are safe. Avoiding activities that cause chest pain or shortness of breath or could result in injuries and bleeding is recommended.
- ▶ **Pregnancy:** Pregnancy carries some significant risks for women who have been treated for aplastic anemia. If a woman has had aplastic anemia and is pregnant or wants to get pregnant, find an aplastic anemia specialist and an obstetrician who specializes in high-risk births.
- ▶ **Surgery:** Surgery can cause serious bleeding in people with a low platelet count. Platelet transfusions may be needed before surgery. The risk for serious infections is also higher. It's a good idea to make sure the aplastic anemia specialist talks with the surgeon prior to having surgery.