Introduction
Aplastic anemia is a rare and serious disease. It happens when your bone marrow fails to make enough blood cells resulting in low blood cell counts. Aplastic anemia can strike people of any age, race, and gender. But, it is more common among children, teenagers, and young adults.

Fortunately, good treatments are currently available. They are helping people with aplastic anemia live longer and resume normal activities.

This reference summary explains what aplastic anemia is. It covers symptoms, causes, diagnosis, and treatment options.

Blood Basics
In order to understand aplastic anemia, it’s important to know certain basic facts about blood.

Blood is made of blood cells floating in a liquid called plasma. Plasma contains proteins, hormones, minerals, and vitamins. It helps move these and the blood cells through your body.
There are three basic types of blood cells:

- **Red blood cells** normally make up almost half of your blood. They are filled with hemoglobin. Hemoglobin is the red part of red blood cells. Its job is to carry oxygen from your lungs and distribute it to your body’s cells.

- **White blood cells** fight disease and infection by attacking and killing germs that get into your body. There are several kinds of white blood cells, each of which fights germs in a different way to stop infection.

- **Platelets** are small cell fragments that help blood clot and stop bleeding.

All three types of blood cells are made by blood forming cells in the bone marrow called bone marrow stem cells. Bone marrow is a spongy tissue located inside most of our bones.

Bone marrow stem cells make copies of themselves all the time. These blood stem cells eventually become mature blood cells. When blood cells are fully formed and functional, they leave the bone marrow and enter the blood.

**Causes**

Aplastic anemia may be acquired or hereditary.

Acquired aplastic anemia is much more common than hereditary aplastic anemia. It can begin anytime in life. About 75 out of 100 cases of acquired aplastic anemia have no known cause.

The other cases of acquired aplastic anemia can be linked to a specific cause or trigger such as medications, toxins, radiation, and other factors. Types of medications that have been linked to aplastic anemia include anti-inflammatory drugs, anti-seizure medications, sulfa drugs and other antibiotics, and treatments for other autoimmune diseases like rheumatoid arthritis or lupus. Aplastic anemia linked to medications is very rare. Most people taking these drugs will not get aplastic anemia.
Exposure to toxins such as pesticides, arsenic, and benzene have been linked to cases of aplastic anemia. Radiation and chemotherapy used to treat cancer have been linked to some cases of aplastic anemia.

Other factors linked to aplastic anemia include:

- Herbal supplements
- An episode of hepatitis (liver inflammation)
- Pregnancy (sometimes this aplastic anemia improves on its own after the woman gives birth.)

Acquired aplastic anemia is due to the destruction of blood forming stem cells in the bone marrow. Most researchers believe that this stem cell destruction happens because the body’s immune system attacks its own cells by mistake.

Normally, the immune system attacks only foreign substances. When your immune system attacks your own body, you are said to have an autoimmune disease. Aplastic anemia is generally thought to be an autoimmune disease. Other autoimmune diseases include rheumatoid arthritis and lupus.

Acquired aplastic anemia cannot be passed from person to person and is not contagious.

Some people with diseases called inherited bone marrow failure syndromes can develop aplastic anemia over time. These diseases can be passed from parent to child and are sometimes referred to as hereditary aplastic anemia.

This form of bone marrow failure is not an autoimmune disease, but instead, the bone marrow failure occurs as a result of the genetic abnormality in the patient.
Some inherited bone marrow failure syndromes include:
- Diamond-Blackfan anemia
- Dyskeratosis congenita
- Fanconi’s Anemia
- Shwachman-Diamond syndrome

Bone marrow failure syndromes are usually diagnosed in childhood and are much less common than acquired aplastic anemia. Less than 20 out of 100 people with aplastic anemia have an identified hereditary cause.

Symptoms
The course of aplastic anemia varies from person to person. You may have only mild symptoms or you may have more severe symptoms. Symptoms are related to your blood cell counts. The lower the blood cell count, the more likely you are to have symptoms.

A person with aplastic anemia can have any of the following:
- A low red blood cell count is called anemia. Red blood cells carry oxygen from your lungs to the rest of your body.
- A low white blood cell count is called leukopenia. White blood cells fight infections in the body. The type of white cells which are most important for attacking and killing bacteria are called neutrophils. A low neutrophil count is called neutropenia.
- A low platelet count is called thrombocytopenia. Platelets help blood to clot and stop bleeding.

The symptoms of low blood cell counts depend on which type of blood cell is affected and how low the counts are.

In aplastic anemia, all three blood cell types are usually reduced. You may have many symptoms, or just one or two. And you may get a new symptom at any point in the course of your illness.
If you have a low red blood cell count, your symptoms may include:

- Tiredness
- Feeling less alert or having trouble concentrating
- Pale skin
- Trouble breathing
- Rapid heartbeat
- Chest pain or pressure

If you have a low white blood cell count, you are more likely to develop infections. Symptoms may include:

- A fever
- Bladder infections that may make it painful to urinate or make you urinate more often
- Lung infections that may make breathing hard

Other symptoms of a low white blood cell count may include:

- Mouth sores
- Sinus infections and a stuffy nose
- Skin infections

If you have a low platelet count, your symptoms may include:

- Bruising or bleeding more easily, even from minor scrapes and bumps
- Nose bleeds

A low platelet count can also cause tiny, flat red spots under your skin, which are caused by bleeding. These spots are called petechiae. These occur more often on the lower legs and belly area.

Bleeding gums, even from simply brushing your teeth, can be a symptom of a low platelet count. Be sure to check with your doctor before any dental work.
Diagnosis

Aplastic anemia is a complex disease. Diagnosing it can be a complex process. Doctors use a number of tools and tests to help them understand your aplastic anemia better.

To understand what is causing your symptoms and low blood counts, your doctor will take a detailed medical history. Your doctor may ask you questions about your symptoms, past medical treatments, and exposure to medicines including herbal supplements, known toxins, or harmful chemicals.

When trying to figure out the cause of your symptoms, your doctor will ask for blood and bone marrow samples. A bone marrow sample, also called a bone marrow aspirate and biopsy, is required for a diagnosis of aplastic anemia. The blood and bone marrow samples are used in a number of tests.

Your doctor will order a CBC, also known as a complete blood count test. This gives information about the quantity and quality of each type of cell in the blood. Your doctor may check your blood iron level. A shortage of iron can cause anemia.

A test to measure the amount of the hormone erythropoietin, or EPO, in the blood is sometimes done. EPO is a hormone made in the kidneys. It stimulates the formation of red blood cells in the bone marrow. A low EPO level may be a sign of a problem other than aplastic anemia.

Vitamin B12 and folate help make blood cells. Checking these levels will tell the doctor how much of these vitamins are in your blood. A shortage of these vitamins can reduce blood cell production and cause a drop in the number of white blood cells, red blood cells, and platelets in the blood.

A reticulocyte count measures the number of young red blood cells in your blood. This test shows whether your bone marrow is making red blood cells at the correct rate. People who have aplastic anemia often have low reticulocyte levels.

Your doctor will look at your bone marrow cells under a microscope to see if they look unusual.
Your doctor will check the percentage of your bone marrow filled with blood cells. This is known as your bone marrow cellularity. Normal bone marrow has a cellularity of 100 minus your age. If you have aplastic anemia your bone marrow cellularity will be abnormally low.

It is also necessary to check the genes in your bone marrow cells using cytogenetic testing. This lets doctors look at the chromosomes or DNA of your bone marrow cells. Doctors use this test to see if you have any chromosome abnormalities or changes that could be causing the symptoms.

Other genetic testing may be necessary to rule out the presence of an inherited bone marrow failure syndrome. This testing helps your doctor know whether your aplastic anemia is acquired or hereditary.

It is important to know whether your aplastic anemia is acquired or hereditary before starting treatment because different treatments work best for acquired and hereditary aplastic anemia. This will help your doctor choose the best treatment for you.

Once diagnosed, doctors divide aplastic anemia into three groups:

- Moderate
- Severe
- Very severe

If you have moderate aplastic anemia:

- You may have low blood cells counts, but not as low as with severe aplastic anemia
- You may have few or no symptoms
- Your doctor may not recommend treatment. Instead, your doctor may just keep an eye on your blood counts
- Your condition may stay the same for many years

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If you have severe aplastic anemia, at least two of the following are true:

- Your neutrophil count is less than 500 cells per millimeter (<500/mm³)
- Your platelet count is less than 20,000 cells per millimeter (<20,000/mm³)
- You have anemia and your reticulocyte count is less than 20,000 cells per millimeter (<20,000/mm³)

If you have very severe aplastic anemia:

- Your neutrophil count is less than 200 cells per millimeter (<200/mm³)
- Your blood counts are otherwise like those of someone with severe aplastic anemia

Aplastic anemia is related to two other diseases. These include:

- Paroxysmal nocturnal hemoglobinuria (PNH)
- Myelodysplastic syndromes (MDS).

In PNH, an abnormal bone marrow stem cell makes abnormal red blood cells. These PNH red blood cells are attacked by blood proteins and destroyed in the blood stream.

More than 10 out of every 100 people with aplastic anemia will also have at least a small percentage of PNH cells at the time of diagnosis, or will develop some later. Patients can have a mix of these two diseases, with features of both to varying degrees.

In MDS, an abnormal bone marrow stem cell makes abnormal blood cells. A small number of people with aplastic anemia go on to develop MDS. A bone marrow biopsy is necessary to tell if a patient has aplastic anemia or MDS.

**Treatment**

Many years ago, there were no treatments for aplastic anemia. It was considered a fatal disease. Today, with standard treatments, as many as 8 out of 10 patients can get better.
The main goal of aplastic anemia treatment is to increase the number of healthy cells in your blood. When your blood counts go up:

- You are less likely to need a blood transfusion
- You can do more of your regular activities
- Your symptoms are not as bad

Your doctor will look at several issues when finding the best treatment plan for you. These include:

- Your symptoms
- Your age
- How severe your case is

Your doctor will also look at:

- Other conditions or diseases you have, including possible inherited bone marrow failure syndromes
- If someone (often a family member) is willing and able to donate matching bone marrow to you

Treatments for aplastic anemia can be divided into three types:

- Supportive care, which helps the symptoms of the disease while waiting for other treatments to work
- Immunosuppressive drug therapy, which prevents your immune system from attacking your bone marrow stem cells
- Bone marrow transplantation (BMT). Bone marrow transplantation replaces your bone marrow stem cells with healthy cells from a donor.

Supportive care treatments may include:

- Blood transfusions, which involve putting blood cells from a donor into your bloodstream. These temporarily increase blood cell counts until other treatments can work.
- Antibiotics may be used to prevent and treat infection.
• Growth factors may be helpful for patients whose blood counts are not too low. Growth factors are man-made versions of naturally occurring proteins in your body. These proteins tell your bone marrow to make more of one type of blood cell. Red cell, white cell, and platelet growth factors are now available.

• Iron chelation therapy may be used in some patients to reduce high levels of iron in your blood. High levels of iron may happen after receiving lots of red blood cell transfusions.

Immunosuppressive drug therapy may keep your immune system from attacking your own bone marrow. This lets bone marrow stem cells grow back and raises blood counts. The standard immunosuppressive drug therapy used to treat aplastic anemia is anti-thymocyte globulin, or ATG for short. This is typically used along with cyclosporine. ATG can be derived from a horse or a rabbit.

Recent studies have shown that horse ATG is more effective than rabbit ATG for many patients with aplastic anemia. This therapy improves blood counts in between 6 and 7 out of 10 cases. Response rates may be even higher in younger patients.

Bone marrow transplantation is the only possible cure for aplastic anemia. Bone marrow transplantation is a two-step process. The first step in a bone marrow transplantation is to destroy existing bone marrow using chemotherapy and sometimes radiation. During the next step of a bone marrow transplantation, the doctor puts blood-forming stem cells from a healthy donor into your bloodstream. These cells travel to your bone marrow and start making healthy blood cells. This process is called engraftment and usually takes 2 to 4 weeks.
Bone marrow transplantation is typically the treatment of choice for aplastic anemia patients under age 40 who have a matched related donor.

If a matched related donor is not available, your doctor can help search for a matched unrelated donor. Success rates with matched unrelated donors have been improving. Therefore, if a well-matched unrelated donor is available for a younger patient, typically under age 40, this is an important option to consider.

Bone marrow transplantation is a complex process and carries serious risks. Some of these include:

- Life-threatening infections
- Graft rejection, where the donor stem cells do not grow in the patient's bone marrow

Bone marrow transplantation also carries the risk of a serious and sometimes chronic immune response called graft-versus-host disease, or GVHD.

When you receive a bone marrow transplant you also get a new immune system from the donor. GVHD happens when the donor’s immune system starts attacking the transplant recipient’s body. GVHD can range from mild to very severe.

If you and your doctor think you may be a candidate for bone marrow transplantation, you should start looking for a donor as soon as possible. It can take a long time to find a matched donor.

If immunosuppressive drug therapy or bone marrow transplantation are not available to you or do not work well, other options can be explored. Many of these options are experimental and are available only through clinical trials.

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Here are 10 good questions to ask your doctor about your treatment:

1. What are all my treatment options?
2. What treatment option do you recommend for me? Why?
3. How likely am I to get better with the treatment?
4. Has this treatment been used a lot? Or, is it a new or experimental treatment?
5. How long will the treatment take to work? When will I know if it is working?
6. Can my disease return, even after successful treatment?
7. How do I take the medicine? How often do I need to take it?
8. How long will I need to take the medicine?
9. What are the common side effects of this medicine or treatment? What can be done to control them?
10. What are the long-term side effects of this medicine or treatment?

Living Well with Aplastic Anemia

In addition to the medical treatments you get, your lifestyle plays a key role in managing your aplastic anemia. By taking good care of your body and mind, and taking an active role in your care, you allow yourself to be as healthy as possible. Here are some tips:

Eat a Healthy and Well-Balanced Diet

Although there is no specific diet that is best for people with aplastic anemia, getting proper nutrition is important for healthy blood production.

Because your nutritional needs can be affected by your disease and medications you take, you should check with your doctor about your individual nutritional needs and the best diet for you. Consider working with a registered dietician to make sure you get the correct nutrition as you go through treatment.
Get the Right Amount of Exercise

It's good for your body to get some form of regular exercise. But if you have aplastic anemia, you may not have much energy to stay active. Plus, you may need to take special precautions. Your doctor can help you find the best fitness plan for you.

If you have a low platelet count:
- You should avoid any activities that could cause bleeding.
- Ask your doctor about safe ways to stay active.
- Add an exercise physiologist to your healthcare team for personalized advice about safe, helpful exercise.
- Tell your doctor if you get a bad headache, dark bowel movements, or severe joint pain. This could mean you are bleeding in the brain, stomach or intestines or the joints.

If you have a low red cell count:
- You should ask your doctor what amount of exercise and what activities will work for you.
- Tell your doctor if you have chest pains or shortness of breath when you exert yourself.
- Tell your doctor if you are going on an airplane or traveling in the mountains. You may need a red blood cell transfusion to make sure you get enough oxygen.

Protect Yourself from Getting Infected by Germs
- Brush your teeth often with a soft brush to prevent the need for dental work. Dental work can cause infection. Check with your doctor before flossing.
- Keep minor infections from getting serious. Tell your doctor if you have a fever or feel very tired. These can be early signs of infection.
- Make sure your food is not too hot. Burns can lead to infections in your mouth.
If you have a very low white count you should take these extra steps to protect yourself from infection:

- Wash your hands often and avoid touching door handles or other things which many people touch.
- Carry a lotion that kills bacteria (anti-bacterial) when you go out. Use it if you can’t find soap and water.
- Stay away from crowds and sick people.

Relax and Reduce Stress

Different people react in different ways to having aplastic anemia. Here are some other ways to help manage your outlook:

- Join a support group.
- Learn more about aplastic anemia.
- Speak with a chronic illness counselor.
- Consider medicine if your doctor says it will help you manage your stress and anxiety.

Summary

Aplastic anemia happens when your bone marrow stops making enough blood cells. Healthy bone marrow stem cells are required for blood cell production. When they are killed or don’t grow right, your blood counts fall, often to very low levels.

There are two types of aplastic anemia – acquired and hereditary. Acquired aplastic anemia happens when your immune system attacks and kills your bone marrow stem cells. Hereditary aplastic anemia is caused by an inherited genetic change in bone marrow stem cells causing them to stop growing right.

People with aplastic anemia may have:

- A low red cell count that can cause drowsiness and low energy.
- A low white cell count that makes it difficult to fight infections.
- A low platelet count that makes it easier to bruise or bleed.
In order to diagnose aplastic anemia, your doctor will take samples of your blood and bone marrow for tests.

Thanks to advances in medicine, many options are available to treat aplastic anemia. They include:

- Supportive care, such as red blood cell and platelet transfusions, antibiotics, and growth factors
- Immunosuppressive drug therapy
- Bone marrow transplantation

Taking an active role in your care is important for staying healthy and getting your best treatment. Here are six tips:

1. Find an expert on aplastic anemia who you can work with.
2. Learn all you can about aplastic anemia and possible treatment options.
3. Make the most of every doctor's visit by bringing a list of questions and getting answers you understand.
4. Keep careful track of all your medical information and records.
5. Work with your doctor to make a treatment plan you are comfortable with.
6. Put together a strong personal support team. Ask for help when you need it.

Beyond choosing and sticking with your medical treatments, there are things you can do to stay well. By eating a healthy and balanced diet, choosing the right exercise, preventing infections, and taking time to relax you allow yourself to be as healthy as possible in order to fight and treat this disease.

Doctors and scientists continue to make great strides in treating aplastic anemia and its symptoms. Learning about aplastic anemia will empower you to cope with this disease. Your doctor and healthcare providers can give you additional information.