CONDITION

Aplastic anemia

Learn how aplastic anemia affects blood cell production, what symptoms may occur, how doctors diagnose it, and which treatments and follow-up care may help.

WHAT IT IS
Bone marrow disorder causing low blood cell production

BODY SYSTEM
Blood and bone marrow

MAIN SYMPTOMS
Tiredness, recurrent infections, easy bruising, unusual bleeding

WHAT CAUSES IT
Abnormal immune response, inherited disorders, often unknown

HOW IT IS DIAGNOSED
Blood counts and bone marrow aspiration and biopsy

MAIN TREATMENT
Stem cell transplantation or immunosuppressive therapy

CAN IT BE CURED?
Sometimes - stem cell transplant may cure

IS IT AN EMERGENCY?
Sometimes - fever with low neutrophils or uncontrolled bleeding

LONG-TERM OUTLOOK
Variable, with possible relapse or clonal evolution

Overview

Aplastic anemia is a disorder in which the bone marrow does not make enough new blood cells. Bone marrow is the soft tissue inside certain bones where blood cells are produced.

The condition can lower one or more of the three main types of blood cells:

When all three types are low, the medical term is pancytopenia . In aplastic anemia, a bone marrow sample usually contains fewer blood-forming cells than expected. Much of the marrow space may be replaced by fat.

Aplastic anemia is not the same as common iron-deficiency anemia. It is also not itself a cancer, although some bone marrow cancers can cause similar blood counts and must be ruled out.

The condition matters because low blood counts can lead to serious infections, bleeding, fatigue, and strain on the heart and other organs. Severity varies. Some people have stable, moderately low counts, while others develop very low counts that require prompt treatment.

Aplastic anemia may develop at any age. Care is usually coordinated by a hematologist, a doctor who specializes in blood disorders. Evaluation at a center experienced in bone marrow failure may be helpful, especially when the condition is severe or a stem cell transplant is being considered.

- Red blood cells , which carry oxygen
- White blood cells , which help fight infection
- Platelets , which help stop bleeding

Symptoms

Symptoms depend on which blood cells are low and how quickly the counts fall. Mild or slowly developing aplastic anemia may first be found during a routine blood test.

Too few red blood cells can cause:

A shortage of neutrophils, an important type of white blood cell, makes it harder to fight bacterial and fungal infections. This may cause fever, chills, mouth sores, sore throat, cough, or infections that keep returning.

Low platelets may lead to easy bruising, tiny red or purple skin spots called petechiae , nosebleeds, bleeding gums, or bleeding that lasts longer than expected. Menstrual bleeding may become heavier.

People with low blood counts should follow the urgent-care instructions provided by their treatment team. A fever can be an urgent problem when neutrophil levels are low, even if other symptoms seem mild. Contact the care team promptly for a new fever, signs of infection, or unusual bleeding.

Call emergency services for severe trouble breathing, chest pain, fainting, confusion, bleeding that will not stop, vomiting blood, black or bloody stools, or symptoms of a stroke. These are emergency symptoms rather than issues to wait and discuss at a routine visit.

- Tiredness or weakness
- Shortness of breath with activity
- Dizziness or headaches
- Pale skin

- A fast or pounding heartbeat

Causes and risk factors

Most acquired aplastic anemia is thought to result from an abnormal immune response. Immune cells attack the bone marrow's blood-forming stem cells, leaving too few cells available to produce healthy blood.

In many cases, doctors cannot identify what started this immune response. This is sometimes called idiopathic aplastic anemia , meaning that the cause is unknown.

Possible triggers or associations include:

An association does not always prove that a medicine or infection caused the condition. People should not stop a prescribed medicine on their own. A clinician can review the timing, possible alternatives, and the risks of stopping treatment.

Some people have an inherited condition that affects bone marrow function. Examples include Fanconi anemia and disorders involving telomeres, the protective ends of chromosomes. An inherited disorder may not become apparent until adolescence or adulthood. Physical features, family history, early graying, lung or liver disease, or unusual reactions to cancer treatment may provide clues, but some people have no obvious signs.

Aplastic anemia can also overlap with paroxysmal nocturnal hemoglobinuria , or PNH. In PNH, some blood cells lack protective surface proteins and may be destroyed too early or contribute to blood clots. A small PNH cell population can occur in people with aplastic anemia without causing typical PNH symptoms.

- Certain medicines, including some cancer treatments
- Exposure to high-dose radiation
- Certain chemicals, such as benzene
- Hepatitis-associated illness and some other viral infections
- Pregnancy, in uncommon cases
- Other autoimmune conditions

Diagnosis

Diagnosis begins with a medical history, physical examination, and blood testing. The clinician may ask about infections, medicines, chemical exposure, radiation, pregnancy, other health

conditions, and blood disorders in the family.

A complete blood count measures red cells, white cells, and platelets. A reticulocyte count measures young red blood cells. A low reticulocyte count can show that the marrow is not producing red cells normally. A blood smear allows specialists to examine the size and shape of blood cells under a microscope.

A bone marrow aspiration and biopsy are central to diagnosis. During this procedure, a clinician removes liquid marrow and a small piece of bone, usually from the back of the hip. Local anesthetic is used to numb the area. The samples show how many blood-forming cells are present and whether abnormal or cancerous cells can be seen.

Doctors also test for other conditions that can resemble aplastic anemia. These include myelodysplastic syndromes, leukemia, vitamin deficiencies, infections, immune disorders, and inherited bone marrow failure syndromes.

Additional testing may include:

Aplastic anemia is classified as nonsevere, severe, or very severe using blood counts and bone marrow findings. This classification helps guide treatment, but doctors also consider symptoms, transfusion needs, age, overall health, other illnesses, and whether a suitable stem cell donor is available.

- Liver, kidney, vitamin, and infection-related blood tests
- Flow cytometry to look for a PNH cell population
- Chromosome or genetic testing on blood or bone marrow cells
- Testing for inherited bone marrow failure when the history or treatment plan makes this relevant
- Tissue typing to identify possible stem cell donors

Treatment and management

Treatment aims to restore blood cell production, prevent infection and bleeding, and reduce symptoms. The plan depends on the severity of the disease and the person's overall situation.

Some people with nonsevere disease and stable counts may be monitored closely rather than treated immediately. Treatment may become appropriate if counts fall, symptoms develop, or regular transfusions are needed.

Supportive care helps manage low blood counts while the marrow recovers or other treatment takes effect. It may include red blood cell transfusions for significant anemia and platelet transfusions for bleeding or very low platelet levels. Transfusion decisions are based on more

than a single laboratory number.

Antibiotics like amoxicillin or doxycycline , antiviral medicines, or antifungal medicines may be used to prevent or treat infection in selected situations. Fever during severe neutropenia needs prompt medical assessment because infections can progress quickly.

Two main treatments can address the underlying marrow failure:

Stem cell transplantation replaces the unhealthy blood-forming system with stem cells from a donor. It is also called an allogeneic hematopoietic stem cell transplant. Before the transplant, treatment is given to prepare the body and suppress the immune system. The donated cells then travel to the bone marrow and begin producing blood cells.

Transplantation can provide long-term disease control and may be curative. Whether it is the preferred option depends on age, health, disease severity, donor availability, and expected treatment risks. Important risks include infection, organ injury, infertility, rejection of the donated cells, and graft-versus-host disease, in which donor immune cells attack the recipient's tissues.

Immunosuppressive therapy reduces the immune attack on bone marrow stem cells. Standard treatment for severe acquired aplastic anemia commonly includes antithymocyte globulin, or ATG, together with cyclosporine. Eltrombopag, a medicine that encourages blood cell production, is often added in appropriate patients.

These medicines require specialist supervision. They can affect the immune system, liver, kidneys, blood pressure, or other organs. Blood tests and clinical monitoring are needed during treatment. Improvement may take months, and not everyone responds fully.

Even after a response, aplastic anemia can return. Some people need additional immunosuppressive therapy or a transplant if the first treatment does not work or loses its effect. Medicines called growth factors have a limited role but may be used in selected clinical situations.

Patients should discuss over-the-counter medicines, supplements, dental work, vaccines, travel, and planned procedures with their care team. When platelets are low, medicines that affect clotting may increase bleeding risk. However, prescribed medicines should not be stopped without medical advice.

Complications

Possible complications come from the low blood counts, the underlying marrow disorder, or its treatment.

Severe infections can occur when neutrophils are very low. Low platelets increase the risk of significant bleeding. Long-lasting anemia may reduce exercise tolerance and place added strain on the heart.

Repeated red blood cell transfusions can cause excess iron to build up in organs. Clinicians may monitor iron levels and discuss treatment if iron overload becomes a concern.

Some people later develop PNH, myelodysplastic syndrome, or acute myeloid leukemia. This process is called clonal evolution , meaning that a group of abnormal marrow cells begins to grow. It does not happen to everyone, but it is one reason long-term monitoring remains important.

Treatment can also cause complications. Immunosuppressive therapy may increase infection risk or affect organ function. Transplantation has its own short- and long-term risks, including graft-versus-host disease and effects on fertility or hormone function.

Follow up

Follow-up is usually more frequent during diagnosis and active treatment. It may become less frequent after blood counts stabilize, but long-term hematology care is still important.

Visits may include a complete blood count, reticulocyte count, medicine review, and tests of kidney or liver function. Doctors may periodically check for a PNH cell population or signs of abnormal marrow cell growth. A repeat bone marrow biopsy may be recommended if counts worsen, new cell abnormalities appear, or the treatment response is unclear.

Routine follow-up is the right time to discuss fatigue, menstrual bleeding, bruising, medication effects, emotional health, fertility, work or school needs, and infection precautions. Patients who have received many transfusions may also need monitoring for iron overload.

Between visits, contact the treatment team promptly for a fever, new signs of infection, worsening shortness of breath, or unusual bleeding. Severe breathing difficulty, chest pain, fainting, confusion, neurologic symptoms, or uncontrolled bleeding require emergency care.

Sources

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