CONDITION

Fanconi anemia

Fanconi anemia is an inherited condition that impairs DNA repair, causing bone marrow failure and a higher cancer risk.

WHAT IT IS
Inherited DNA-repair and bone marrow failure condition

BODY SYSTEM
Blood, bone marrow, development, and multiple organs

MAIN SYMPTOMS
Fatigue, frequent infections, easy bruising, growth differences

WHAT CAUSES IT
Inherited changes in DNA-repair genes

HOW IT IS DIAGNOSED
Chromosome breakage testing and genetic testing

MAIN TREATMENT
Supportive care, monitoring, and stem cell transplant

CAN IT BE CURED?
No - transplant treats marrow failure, not inherited risks

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

LONG-TERM OUTLOOK
Lifelong specialist care with serious blood and cancer risks

Overview

Fanconi anemia is a rare inherited condition that affects how cells repair damaged DNA. When repair does not happen properly, cells can become damaged or die.

The bone marrow, which makes red blood cells, white blood cells, and platelets, is especially vulnerable. In Fanconi anemia, production of these cells often falls over time. This is called bone marrow failure.

Low red blood cells can cause anemia and tiredness. Low white blood cells can make infections harder to fight. Low platelets can cause easy bruising or bleeding.

Fanconi anemia can also affect growth, development, hormones, fertility, and organs such as the kidneys or heart. Some people have noticeable differences from birth, while others do not.

The condition raises the lifetime risk of myelodysplastic syndrome, leukemia, and certain solid cancers.

Despite its name, Fanconi anemia is not only a type of anemia. It affects DNA repair throughout the body and is different from Fanconi syndrome, a kidney disorder.

Symptoms

Signs differ greatly between people, including members of the same family. Some are identified at birth because of physical differences. Others are diagnosed when blood counts fall, growth is slower than expected, or cancer develops unusually early.

Possible physical and developmental features include:

Not everyone has these features. Fanconi anemia can occur without obvious physical differences.

Anemia can cause tiredness, weakness, pale skin, headaches, shortness of breath, or reduced stamina. A shortage of neutrophils, a type of white blood cell, may lead to frequent or serious infections. Low platelets can cause easy bruising, small red or purple skin spots, nosebleeds, bleeding gums, or prolonged bleeding.

Symptoms cannot confirm Fanconi anemia because many more common conditions cause similar problems.

- Shorter height or slower growth
- Smaller head size
- Differences affecting the thumbs, hands, or forearms
- Lighter or darker skin areas, including café-au-lait spots
- Kidney, urinary tract, heart, hearing, or eye differences
- Delayed puberty or other hormone-related concerns
- Learning or developmental difficulties

Causes and risk factors

Fanconi anemia is caused by a harmful change in one of several genes involved in DNA repair.

Most forms follow an autosomal recessive inheritance pattern. A child inherits one altered copy of the same gene from each parent, who are usually healthy carriers.

Less commonly, Fanconi anemia is X-linked and involves the FANCB gene. Rare forms follow a dominant pattern, in which one altered gene copy can cause the condition.

The specific gene change may influence symptoms and cancer risks but cannot predict the exact course. Genetic counseling can explain the inheritance pattern and testing options for relatives.

Fanconi anemia is not caused by diet, stress, infection, something done during pregnancy, or contact with another person.

Diagnosis

Fanconi anemia may be suspected because of physical features, persistently low blood counts, bone marrow failure at a young age, or family history. Some people are first assessed after developing myelodysplastic syndrome or cancer unusually early.

A chromosome breakage test is central to diagnosis. Blood cells are exposed in a laboratory to diepoxybutane, often called DEB, or mitomycin C. Cells affected by Fanconi anemia show a characteristic increase in chromosome breaks and rearrangements.

Sometimes blood-forming cells naturally correct the gene problem through somatic mosaicism. This can make a blood test unclear or negative. If suspicion remains, testing may be repeated using skin cells called fibroblasts.

Genetic testing can identify the affected gene and specific changes. This may confirm the diagnosis, clarify inheritance, guide family testing, and support transplant planning. A negative or uncertain genetic result does not always exclude Fanconi anemia if chromosome breakage testing is clearly abnormal.

Other assessments may include:

Testing should be coordinated by a team experienced in inherited bone marrow failure conditions.

- Complete blood counts and bone marrow examinations
- Kidney, heart, hearing, vision, growth, and hormone assessments
- Tissue-matching tests if stem cell transplant is being considered
- Age-appropriate examinations for associated cancers

Treatment and management

Treatment depends on blood counts, symptoms, complications, age, and overall health. Care may involve blood and cancer specialists, transplant teams, genetic counselors, and clinicians experienced in hormone, kidney, hearing, fertility, and developmental concerns.

Regular blood counts and periodic bone marrow examinations help track marrow function and detect worsening failure, myelodysplastic syndrome, or leukemia.

Red blood cell or platelet transfusions can treat severe anemia or bleeding risk, but they do not correct the underlying condition. Clinicians consider the effects of repeated transfusions when planning possible transplant care.

Androgen medicines can improve blood counts in some people. Their effect may be temporary, and they can cause significant liver, hormone, growth, and other side effects. They require specialist supervision and monitoring.

A hematopoietic stem cell transplant, also called a bone marrow transplant, replaces unhealthy blood-forming cells with donor cells. It can cure bone marrow failure and treat some blood cancers, but it does not correct physical differences or remove the increased risk of solid cancers.

People with Fanconi anemia are unusually sensitive to some chemotherapy medicines and radiation because their cells cannot repair DNA damage normally. Transplant preparation and cancer treatment require specialist protocols. Standard treatment designed for people without Fanconi anemia may cause excessive toxicity.

Supportive care may include treatment for infections, hormone problems, hearing loss, nutrition concerns, or developmental needs. Fertility preservation can be discussed before treatments that may further reduce fertility.

Gene therapy is being studied but is not an established routine treatment.

Complications

Progressive bone marrow failure is a major complication. When red cells, white cells, and platelets are all low, this is called pancytopenia. It can cause severe fatigue, infections, and bleeding.

Damaged marrow cells may develop into myelodysplastic syndrome, in which abnormal blood-forming cells do not mature properly. Some cases progress to acute myeloid leukemia.

Fanconi anemia also raises the risk of solid cancers, particularly squamous cell cancers of the head and neck, mouth, throat, esophagus, and genital or anal areas. These may occur at younger ages than in the general population, and the risk remains after a successful stem cell

transplant.

Other complications include reduced fertility, early menopause, delayed puberty, thyroid or blood sugar problems, low bone density, and liver disease. Kidney, heart, hearing, or skeletal differences may also need continuing care.

These risks make lifelong monitoring important even when blood counts are stable.

Prevention

Fanconi anemia cannot currently be prevented in someone who has inherited the gene changes that cause it.

Genetic counseling can explain the chance of passing the condition to a child. Depending on the family's goals, options may include carrier testing, testing during pregnancy, or testing embryos created through in vitro fertilization. These are personal choices, not required steps.

Avoiding tobacco and limiting alcohol can reduce added cancer risk. Sun protection lowers avoidable ultraviolet damage. Vaccination, including protection against human papillomavirus when appropriate, can help prevent some cancer-linked infections.

Vaccination and screening plans should account for age, immune function, transplant history, and local recommendations.

Living with it and what to expect

Fanconi anemia is lifelong, but its course varies. Blood counts may remain stable and then decline over months or years. Physical features present from birth generally remain, while growth, hormone, or fertility concerns may become more noticeable with age.

Some children need frequent medical care early in life. Others attend school and take part in everyday activities with adjustments for fatigue, infections, appointments, or physical differences. Adults may need support with work, fertility decisions, and cancer screening.

A stem cell transplant can restore blood production, but recovery takes time. Infection precautions, medicines, and frequent follow-up are often needed during the first months. Later follow-up checks for transplant effects, organ health, hormone concerns, and cancer. Transplant does not end the need for lifelong care.

Routine follow-up usually includes blood monitoring and examinations selected for the person's age, gene type, treatment history, and health. Keep a clear record of the diagnosis because emergency, surgical, dental, and cancer teams need to know about sensitivity to

radiation and certain medicines.

Uncertainty about blood counts, cancer risk, appearance, fertility, or transplant can affect the whole family. Counseling, school support, and experienced patient organizations may help.

When to get medical help

Contact the usual care team promptly for new tiredness, increasing bruising, repeated nosebleeds, bleeding gums, frequent infections, unexplained weight loss, persistent mouth sores, a new lump, or a sore throat or hoarse voice that does not settle. These symptoms often have other causes but should not be ignored.

Keep routine appointments even when you feel well. Falling blood counts and early tissue changes may not cause obvious symptoms.

Seek urgent medical advice for a fever if you have low neutrophils, are receiving cancer treatment, or recently had a stem cell transplant. Follow the fever plan provided by your specialist team.

Get emergency help for:

Do not delay emergency care while waiting to contact the specialist team.

- Bleeding that is heavy or will not stop
- Severe trouble breathing
- Fainting, confusion, or difficulty waking
- Sudden severe weakness
- Signs of a serious allergic reaction

Common questions

Is Fanconi anemia a type of cancer?

No. It is an inherited DNA-repair condition, but it substantially increases the risk of myelodysplastic syndrome, leukemia, and several solid cancers.

Can Fanconi anemia be cured with a bone marrow transplant?

A transplant can cure bone marrow failure by replacing the blood-forming system. It does not remove the underlying gene change from the rest of the body, so cancer and other health risks remain.

Is Fanconi anemia contagious?

No. It cannot spread through touch, coughing, blood, food, sex, or shared living spaces. It is caused by inherited gene changes.

Can someone have Fanconi anemia without birth differences?

Yes. Some people have no obvious physical differences and are diagnosed because of low blood counts, cancer, chromosome breakage testing, or testing after a relative is diagnosed.

Is Fanconi anemia the same as Fanconi syndrome?

No. Fanconi anemia is an inherited DNA-repair and bone marrow failure condition. Fanconi syndrome affects the kidney tubules and causes important substances to be lost in urine.

Can a person with Fanconi anemia have children?

Some can, but reduced fertility is common in all sexes. Pregnancy may require specialist planning because of blood counts and other health risks. Genetic counseling can explain the chance of passing on the condition.

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