CONDITION

DBA

DBA is an inherited bone marrow condition that mainly reduces red blood cell production.

WHAT IT IS
Inherited red blood cell production disorder

BODY SYSTEM
Blood and bone marrow

MAIN SYMPTOMS
Pale skin, tiredness, breathlessness, poor growth

WHAT CAUSES IT
Usually inherited or new gene changes

HOW IT IS DIAGNOSED
Blood tests, bone marrow findings, often genetic testing

MAIN TREATMENT
Transfusions, corticosteroids, or stem cell transplant

CAN IT BE CURED?
Sometimes - stem cell transplant can cure blood problems

IS IT AN EMERGENCY?
Sometimes - severe breathlessness, chest pain, or fainting

LONG-TERM OUTLOOK
Many need lifelong monitoring; treatment needs differ

Overview

DBA stands for Diamond-Blackfan anemia. It is a rare condition in which the bone marrow does not make enough red blood cells. Bone marrow is the soft tissue inside bones where blood cells are produced.

Red blood cells carry oxygen around the body. Having too few healthy red blood cells causes anemia, which can leave the body's tissues without enough oxygen.

DBA usually becomes apparent during infancy because a baby is pale, feeds poorly, grows slowly, or has a low red blood cell count. Less often, it is recognized later in childhood or adulthood. Mild forms may be found only after a blood test or genetic assessment.

DBA is an inherited bone marrow failure syndrome. Although it mainly affects red blood cells, it can also be linked with physical differences present from birth, growth concerns, and an

increased risk of certain cancers. Lifelong follow-up matters even when anemia is well controlled.

Symptoms

Symptoms mainly result from anemia and may develop gradually. They can include:

A baby may be unusually sleepy or tire during feeding. Older children may struggle with usual play or exercise, while adults may notice reduced stamina.

Some people with DBA have physical differences affecting the thumbs, hands, face, head, heart, kidneys, urinary system, or reproductive organs. Shorter height can also occur. Not everyone has these features, and their absence does not rule out DBA.

Symptoms alone cannot confirm DBA. More common conditions, including iron deficiency and infections, can also cause anemia.

- Pale skin, lips, or nail beds
- Tiredness, weakness, or reduced activity
- Breathlessness, especially during activity or feeding
- A fast heartbeat
- Poor feeding or slow weight gain in a baby
- Headaches, dizziness, or difficulty concentrating

Causes and risk factors

DBA is usually caused by a change in a gene involved in making ribosomes, structures inside cells that help build proteins. Developing red blood cells are especially sensitive when this process does not work normally.

Many known DBA-related genes provide instructions for parts of a ribosome. Others affect red blood cell development in different ways. Genetic testing does not identify a cause in every person with DBA.

A gene change may be inherited from a parent or arise for the first time in the egg, sperm, or early embryo. This is called a new, or de novo, variant.

Many forms follow an autosomal dominant inheritance pattern, meaning one altered gene copy can cause the condition. However, the same genetic change may affect people differently. A parent with few symptoms may have a child with more significant anemia. Some uncommon

forms have other inheritance patterns.

Having a close relative with DBA or a related gene change raises the chance of having it. DBA is not caused by diet, parenting, ordinary infections, or anything done during pregnancy.

Diagnosis

DBA is usually assessed by a pediatric hematologist or another blood specialist. Diagnosis combines medical history, examination, blood tests, bone marrow findings, and sometimes genetic results.

A complete blood count measures the number and size of blood cells. In DBA, red blood cells are often larger than usual. The reticulocyte count is typically low. Reticulocytes are young red blood cells, so a low count suggests that the bone marrow is not producing enough new cells. White blood cells and platelets are often less affected, particularly near diagnosis.

A bone marrow examination may be needed. A small sample, usually from the hip bone, is examined under a microscope. DBA typically causes very few developing red blood cells while other blood cell lines are relatively preserved.

Genetic testing can support diagnosis, clarify inheritance, and guide family testing. A negative result does not always exclude DBA because not every causative change is known or detectable.

Other tests may identify raised fetal hemoglobin or increased red blood cell adenosine deaminase activity. These findings cannot diagnose DBA on their own.

The assessment also rules out nutritional deficiencies, parvovirus infection, temporary childhood red cell suppression, medication effects, and other inherited bone marrow conditions. Imaging or specialist examinations may check the heart, kidneys, bones, hearing, or other areas when appropriate.

Treatment and management

Treatment depends on the severity of anemia, age, symptoms, other health needs, and response to previous treatment. Mild anemia may only require monitoring.

Red blood cell transfusions increase hemoglobin and improve oxygen delivery. They are often used in babies, during serious illness, or when another treatment is unsuitable. Some people need regular transfusions; others need them only at certain times.

Repeated transfusions can cause iron to collect in the liver, heart, and hormone-producing

organs. Blood tests and scans monitor this buildup, and iron chelation medicines may help remove it.

Corticosteroids like prednisone or hydrocortisone can encourage red blood cell production in some people. Specialists generally avoid starting them in young infants because they can affect growth and development.

If corticosteroids work, the care team aims to use the lowest dose that maintains a safe hemoglobin level. Long-term use can affect growth, bones, blood pressure, blood sugar, eyes, infection risk, and appearance, so regular monitoring is important. Some people do not respond or cannot tolerate the side effects.

A hematopoietic stem cell transplant replaces the blood-forming system with healthy donor cells. It can cure the bone marrow and anemia problems caused by DBA and may be considered for severe transfusion dependence, major treatment complications, or serious additional bone marrow problems.

Transplantation has significant risks, including infection, organ injury, and graft-versus-host disease, in which donor immune cells attack the recipient's tissues. An experienced transplant team must assess the benefits and risks. A transplant does not remove the genetic change from the rest of the body or correct existing physical differences.

Other support may include nutrition care, growth and hormone assessment, bone monitoring, psychological support, and planning for school, work, pregnancy, or transition to adult services.

Complications

Poorly controlled anemia can strain the heart and limit growth, activity, learning, or daily functioning. Other possible complications include:

DBA is associated with a higher risk of myelodysplastic syndrome, leukemia, and certain solid tumors. Myelodysplastic syndrome causes the bone marrow to make abnormal blood cells. This increased risk does not mean everyone with DBA will develop cancer.

Cancer risk may remain after a successful stem cell transplant because the inherited change is still present in non-blood cells. Long-term follow-up should include age-appropriate surveillance planned by a team familiar with DBA.

- Iron overload from repeated transfusions
- Reduced growth or weakened bones from long-term corticosteroids
- Transfusion reactions or antibodies against donor blood

- Complications of stem cell transplantation
- Hormone, fertility, heart, or liver problems related to treatment or iron
- Additional changes in blood counts over time

Prevention

DBA caused by an inherited or new gene change cannot usually be prevented and is not caused by lifestyle choices.

If a DBA-related gene change is identified, genetic counseling can explain inheritance and testing options for relatives. It can also provide information about prenatal testing or embryo testing during in vitro fertilization. These are personal reproductive choices, not ways to prevent DBA in someone who already has it.

Regular monitoring can reduce some treatment-related complications. This includes checking for iron overload, reviewing corticosteroid side effects, keeping transfusion records, and attending recommended cancer surveillance.

Living with it and what to expect

DBA varies widely. Some people need regular transfusions throughout life, while others respond to corticosteroids or have a stem cell transplant. A smaller group enters remission, meaning hemoglobin remains at a safe level without transfusions or corticosteroids.

Remission may last for years but is not always permanent. Blood counts and long-term surveillance remain important.

During childhood, care often focuses on growth, development, school attendance, treatment side effects, and physical differences. A written care plan can help schools and caregivers understand fatigue, treatment schedules, and when symptoms need attention.

Adults may need support with work, insurance, fertility, pregnancy planning, and transfer to adult hematology care. Pregnancy places extra demands on red blood cell production, so advance planning with hematology and pregnancy specialists is important.

Follow-up may include blood counts, iron measurements, liver and heart assessment, growth or hormone checks, and bone health reviews. The schedule depends on age, treatment history, genetic findings, and previous complications.

Many people with DBA attend school, work, exercise, travel, and have families. Daily life may still involve medicines, transfusions, appointments, or periods of reduced energy. Emotional

support and contact with others living with DBA can help.

When to get medical help

Arrange a routine appointment for gradually increasing tiredness, paleness, headaches, reduced exercise tolerance, poor feeding, or slower growth. People receiving treatment should report new side effects, missed transfusions, or difficulty taking prescribed medicines.

Seek prompt medical advice for fever or signs of infection if you have had a stem cell transplant, take immune-suppressing medicines, or have a central venous line. Follow any specific instructions from the treating service about temperature thresholds and where to seek care.

Seek emergency help for:

These symptoms can have causes other than DBA, but they need urgent assessment.

- Severe or rapidly worsening difficulty breathing
- Chest pain
- Fainting, severe confusion, or difficulty waking
- Blue or gray lips or skin
- Uncontrolled bleeding
- A severe reaction during a blood transfusion

Common questions

Is DBA a type of cancer?

No. DBA is an inherited bone marrow failure condition, not cancer. However, it increases the lifetime risk of certain blood cancers and solid tumors, so long-term surveillance is recommended.

Can DBA go away on its own?

Some people enter remission and no longer need transfusions or corticosteroids. Remission cannot be predicted and may not be permanent. Follow-up is still needed because other health risks remain.

Is DBA contagious?

No. You cannot catch DBA through contact, coughing, food, blood exposure, or shared

surroundings. It results from a genetic change.

Can adults have DBA?

Yes. DBA usually appears in infancy, but mild cases may not be recognized until later. Adults may be diagnosed after unexplained anemia, physical findings, or genetic testing prompted by a relative's diagnosis.

Can someone with DBA have children?

Many people with DBA can have children. Fertility may be reduced by iron overload, treatment, or physical differences. A DBA-related gene change may be passed on. Genetic counseling and pregnancy planning can clarify individual risks and care needs.

Does a stem cell transplant cure DBA?

A successful transplant can cure bone marrow failure and remove the need for anemia treatment. It does not remove the genetic change from the rest of the body, reverse existing physical differences, or eliminate every long-term cancer risk.

Sources

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