CONDITION

Hereditary elliptocytosis

Hereditary elliptocytosis is an inherited condition that makes many red blood cells oval or elongated.

WHAT IT IS
Inherited oval-shaped red blood cells

BODY SYSTEM
Blood and circulation

MAIN SYMPTOMS
Fatigue, pale skin, jaundice, enlarged spleen

WHAT CAUSES IT
Inherited red blood cell membrane changes

HOW IT IS DIAGNOSED
Blood smear, hemolysis tests, sometimes genetic testing

MAIN TREATMENT
Monitoring and treatment of anemia or complications

CAN IT BE CURED?
No - inherited membrane changes remain

IS IT AN EMERGENCY?
Sometimes - severe anemia or fever after spleen removal

LONG-TERM OUTLOOK
Usually normal lifespan with appropriate monitoring

Overview

Hereditary elliptocytosis is an inherited condition affecting red blood cells. Many of the cells are oval, elongated, or shaped like a narrow ellipse instead of being round with a shallow center.

The shape change happens because proteins supporting the outer membrane of each red blood cell do not work in the usual way. This can make the cells less flexible as they move through small blood vessels and the spleen.

For most people, hereditary elliptocytosis causes few or no health problems. The condition may be found by chance during a blood test. In some people, however, the spleen removes the unusually shaped cells too quickly. This is called hemolysis , meaning red blood cells break down earlier than they should.

Ongoing hemolysis can cause anemia, jaundice, an enlarged spleen, or gallstones. A rare, more severe form called hereditary pyropoikilocytosis can cause marked anemia, particularly during infancy and childhood.

Hereditary elliptocytosis is not an infection or cancer. It cannot spread to another person.

Symptoms

Many people have no symptoms. Their body makes new red blood cells quickly enough to replace those being removed.

When symptoms occur, they usually result from anemia or hemolysis. They may include:

Young children with a more severe form may have poor feeding, slow weight gain, marked paleness, or jaundice. Newborn jaundice needs prompt assessment because very high bilirubin levels can be harmful. Bilirubin is a yellow substance produced when red blood cells break down.

Symptoms can become more noticeable during an infection or another period of physical stress. Pregnancy may also reveal or worsen anemia in some people.

- Tiredness, weakness, or reduced exercise tolerance
- Pale skin or pale tissue inside the lower eyelids
- Yellow skin or eyes, known as jaundice
- Dark urine during periods of increased hemolysis
- Fullness or discomfort under the left ribs from an enlarged spleen
- Gallstone pain in the upper abdomen, often after eating

Causes and risk factors

Hereditary elliptocytosis is caused by changes in genes that provide instructions for proteins in the red blood cell membrane. These proteins help the cells keep their shape and flexibility.

Several genes can be involved, including SPTA1 , SPTB , EPB41 , and GYPC . They affect proteins such as spectrin and protein 4.1, which form part of the cell's internal support structure.

Most forms follow an autosomal dominant inheritance pattern. This means a person can have the condition after inheriting one altered gene copy from either parent. The severity may differ greatly within the same family.

More severe forms, including many cases of hereditary pyropoikilocytosis, may occur when a person inherits particular gene changes from both parents. Genetic testing may help explain the inheritance pattern, but it does not always predict how severe the symptoms will be.

The main risk factor is having a biological parent or close relative with the condition. Sometimes the gene change is discovered before other relatives know they carry it. Less commonly, a new genetic change occurs without a known family history.

Hereditary elliptocytosis is present from birth, even if it is not identified until adulthood. Diet, exercise, stress, or something a parent did during pregnancy does not cause it.

Diagnosis

A clinician may first suspect hereditary elliptocytosis after a complete blood count or examination of a blood smear. A blood smear is a thin layer of blood viewed under a microscope. It can show many oval or elongated red blood cells.

Elliptical cells are not always inherited. Smaller numbers can appear with iron deficiency, thalassemia, certain bone marrow conditions, or other causes of anemia. Diagnosis therefore considers the number and appearance of the cells, your blood results, symptoms, and family history.

Tests may include:

Specialist laboratories may use red cell membrane studies or ektacytometry , which measures how red blood cells change shape under stress. Genetic testing can identify a related gene change in some families. It is especially useful when the blood findings are unclear, symptoms are severe, or family planning information is wanted.

Testing relatives is not always necessary. It may be considered when the result could clarify symptoms, inheritance, or future medical care.

- A complete blood count to check hemoglobin and red blood cell measurements
- A reticulocyte count to measure newly produced red blood cells
- Bilirubin, lactate dehydrogenase, and haptoglobin tests for hemolysis
- A blood smear reviewed by someone experienced in blood cell disorders
- A direct antiglobulin test when immune-related hemolysis needs excluding

Treatment and management

People without anemia or other complications usually need no active treatment. Periodic blood tests may be enough to check that the condition remains stable.

If red blood cells are breaking down quickly, management may include folic acid. The body uses folate, a B vitamin, to make new blood cells. A clinician can advise whether supplementation is appropriate rather than assuming it is needed.

Iron should not be taken solely because anemia is present. Hemolytic anemia is different from iron-deficiency anemia, and unnecessary iron can build up in the body. Iron treatment is used only when testing confirms deficiency.

A blood transfusion may be needed for severe anemia, particularly during infancy, a serious infection, or an aplastic crisis. An aplastic crisis is a sudden pause in red blood cell production, often associated with parvovirus B19 infection.

For severe, ongoing hemolysis, removal of the spleen may be considered. This operation is called a splenectomy . It can reduce anemia because the spleen is a major site where abnormal cells are removed. It does not change the inherited gene or make the cells round.

Splenectomy is generally reserved for people with substantial symptoms or transfusion needs because the spleen helps protect against serious infections. Vaccination, infection precautions, and sometimes preventive antibiotics like amoxicillin or doxycycline are important before and after the operation. The exact plan is based on age and local medical guidance.

Newborn jaundice may be treated with phototherapy, which uses special light to lower bilirubin. More intensive treatment is sometimes required. Symptomatic gallstones may need separate treatment, including possible gallbladder surgery.

Complications

Most people do not develop major complications. When hemolysis is significant, possible problems include:

An enlarged spleen is more vulnerable to injury. Someone with significant spleen enlargement may be advised to avoid activities with a high risk of a blow to the abdomen.

After splenectomy, there is a lifelong increase in the risk of severe bacterial infection. Blood clot risk may also increase in some people. These risks are considered carefully before surgery and require long-term follow-up afterward.

- Persistent anemia
- An enlarged spleen

- Pigment gallstones caused by ongoing bilirubin production
- Sudden worsening of anemia during infection
- Aplastic crisis, particularly with parvovirus B19
- High bilirubin levels in a newborn
- Iron overload after repeated blood transfusions

Prevention

Hereditary elliptocytosis cannot be prevented because it is caused by an inherited genetic change. Lifestyle choices cannot remove that change.

Complications can sometimes be reduced through appropriate monitoring. This may include checking anemia, investigating abdominal pain that could indicate gallstones, and avoiding unnecessary iron treatment.

If you are planning a pregnancy, genetic counselling can explain the chance of passing the condition on and why severity can differ between relatives. Genetic counselling provides information and support; it does not require you to have genetic testing or make a particular reproductive decision.

Living with it and what to expect

For many people, hereditary elliptocytosis remains mild throughout life. The elliptical shape of the red blood cells does not go away, but it may never interfere with everyday activities, education, work, exercise, or pregnancy.

Blood counts often remain fairly stable over months and years. Temporary changes can occur during infections, pregnancy, or other periods when the body needs to make more red blood cells. Infants who have significant hemolysis may improve as they grow, although this cannot be predicted for every child. More severe inherited forms may require continuing specialist care.

Routine follow-up is based on symptoms and severity. It may involve blood counts, checks for hemolysis, and review of spleen size or gallstone symptoms. People receiving repeated transfusions may also need monitoring for excess iron.

Tell healthcare professionals that you have hereditary elliptocytosis when having blood tests or being assessed for anemia. This can help prevent an abnormal blood smear from being mistaken for a new problem.

If your spleen has been removed, keep vaccinations up to date according to your medical plan. You may be advised to carry a medical alert card and follow a specific plan for fever. These precautions remain important even many years after surgery.

When to get medical help

Arrange a routine appointment if you develop new or gradually worsening fatigue, paleness, yellowing of the eyes, dark urine, or discomfort under the left ribs. Repeated upper abdominal pain, especially after meals, may need assessment for gallstones.

Seek prompt medical advice if an infection is followed by a clear increase in weakness, jaundice, breathlessness, or paleness. These symptoms may mean the anemia has worsened.

A newborn with yellow skin or eyes should receive same-day medical assessment. Seek urgent help if the baby is difficult to wake, feeds poorly, has an unusual cry, or appears increasingly yellow.

Get emergency help for severe trouble breathing, chest pain, fainting, confusion, extreme weakness, or rapidly worsening paleness. These can be signs of severe anemia or another urgent condition.

If you have had your spleen removed, fever can signal a serious infection. Follow your individual fever plan and seek urgent medical assessment immediately rather than waiting for a routine appointment.

Common questions

Is hereditary elliptocytosis serious?

It is usually mild, and many people never develop symptoms. A smaller group has ongoing hemolysis, anemia, jaundice, gallstones, or an enlarged spleen. Hereditary pyropoikilocytosis is generally more severe.

Does hereditary elliptocytosis go away?

No. The inherited membrane change and elliptical red blood cell shape remain throughout life. Symptoms can still change over time, and treatment can control anemia or complications when needed.

Is it the same as hereditary spherocytosis?

No. Both are inherited red blood cell membrane conditions, but they produce different cell shapes. Hereditary elliptocytosis causes oval cells, while hereditary spherocytosis causes

round, sphere-shaped cells. Their tests and management overlap in some areas.

Will I pass it to my children?

Many forms are autosomal dominant, so each child can inherit the altered gene from an affected parent. Other inheritance patterns are possible. A genetic counsellor can explain the pattern in your family more precisely.

Will I need my spleen removed?

Most people do not. Splenectomy is considered mainly when ongoing hemolysis causes severe anemia, major symptoms, or repeated transfusion needs. It reduces red blood cell destruction but creates lifelong infection-related precautions.

Can hereditary elliptocytosis turn into leukemia or cancer?

No. Hereditary elliptocytosis does not turn into leukemia or another cancer. However, elliptical cells can also appear in some unrelated blood or bone marrow conditions, which is why new or unexplained blood abnormalities should be assessed properly.

Sources

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