CONDITION

Glanzmann thrombasthenia

Glanzmann thrombasthenia is an inherited condition in which platelets cannot stick together properly to stop bleeding.

WHAT IT IS
Inherited platelet function disorder

BODY SYSTEM
Blood and clotting

MAIN SYMPTOMS
Nosebleeds, easy bruising, gum bleeding, heavy periods

WHAT CAUSES IT
Inherited gene changes affecting platelets

HOW IT IS DIAGNOSED
Platelet aggregation, flow cytometry, and genetic testing

MAIN TREATMENT
Bleeding control, antifibrinolytics, platelets, and clotting factor

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

IS IT AN EMERGENCY?
Sometimes - when bleeding is heavy or will not stop

LONG-TERM OUTLOOK
Lifelong condition often manageable with specialist care

Overview

Glanzmann thrombasthenia is a rare, inherited bleeding condition. It affects platelets, the small blood cells that gather at an injury and help form a plug to stop bleeding.

In this condition, platelets are usually present in normal numbers, but they cannot stick to one another properly. This happens because a protein on their surface, called integrin alpha-IIb beta-3, is missing or does not work as expected. The protein is also known as glycoprotein IIb/IIIa.

Bleeding most often involves the nose, mouth, skin, digestive tract, or menstrual periods. Bleeding after an injury, dental work, surgery, or childbirth can also be significant. Bleeding into joints and muscles, which is typical of haemophilia, is less common.

The condition is lifelong. Its severity differs between people and cannot always be predicted from genetic test results. Specialist planning can help prevent and manage many bleeding problems.

Symptoms

Symptoms often begin in infancy or childhood, although some people are diagnosed later. The type and frequency of bleeding can change over time.

Common features include:

Some babies have prolonged bleeding from the umbilical cord or after circumcision. Bleeding from the digestive tract or urinary tract is less common but can occur.

Blood loss may cause iron-deficiency anaemia. This means the body does not have enough iron to make healthy red blood cells. Possible signs include tiredness, weakness, shortness of breath during activity, headaches, or pale skin.

Symptoms alone cannot confirm Glanzmann thrombasthenia. Several other bleeding conditions can cause similar problems.

- Frequent or prolonged nosebleeds
- Easy bruising or small red or purple skin spots
- Bleeding from the gums or mouth
- Bleeding that continues longer than expected after a cut
- Heavy or prolonged menstrual periods
- Bleeding after losing a tooth, dental treatment, surgery, or injury

Causes and risk factors

Glanzmann thrombasthenia is usually caused by changes in the ITGA2B or ITGB3 gene. These genes provide instructions for making the platelet surface protein needed for platelets to join together.

The condition usually follows an autosomal recessive inheritance pattern. This means a person has inherited one altered copy of the relevant gene from each parent. The parents are usually carriers and do not have the condition themselves.

If two carriers have a child, each pregnancy has:

These chances apply separately to each pregnancy. They do not predict what will happen

across a family.

The condition is more likely in families where both parents carry the same gene change. This can occur in any population, but it is more common in communities where marriage between blood relatives is customary.

Genetic counselling can explain inheritance, carrier testing, and reproductive testing options without directing you toward a particular choice.

- A 1 in 4 chance that the child will have the condition
- A 1 in 2 chance that the child will be a carrier
- A 1 in 4 chance that the child will inherit neither altered copy

Diagnosis

Diagnosis usually involves a haematologist, a doctor who specialises in blood conditions. Your personal and family bleeding history helps guide testing.

A complete blood count usually shows a normal number of platelets. The platelets also often look normal under a microscope. Routine clotting tests, including prothrombin time and activated partial thromboplastin time, are usually normal. Normal results on these tests do not rule out a platelet function problem.

Platelet aggregation testing is central to diagnosis. It measures how platelets respond to substances that normally make them clump together. In Glanzmann thrombasthenia, platelets show little or no aggregation with most of these substances, while their response to ristocetin is generally preserved.

Flow cytometry uses labelled antibodies to measure proteins on the platelet surface. It can show that alpha-IIb beta-3 is missing, reduced, or abnormal.

Genetic testing may identify changes in ITGA2B or ITGB3 . It can confirm the diagnosis, help with family testing, and distinguish the condition from other inherited platelet disorders. In some people, testing does not find a clear genetic explanation even when the platelet results support the diagnosis.

Treatment and management

Care is usually coordinated through a haematology or haemophilia treatment centre. Treatment depends on where the bleeding is, how severe it is, and whether previous platelet transfusions have caused antibodies.

Small cuts may respond to firm, continuous pressure. Nosebleeds and mouth bleeding may need local treatments that compress or seal the bleeding area. Dental teams can use stitches, dressings, or other local measures when appropriate.

Antifibrinolytic medicines help prevent the body from breaking down a newly formed clot too quickly. They are often used for bleeding from the mouth or nose, heavy periods, and planned dental procedures. They may be given by mouth, into a vein, or as a local mouth treatment, depending on the situation.

Platelet transfusions provide functioning platelets and are an important treatment for major bleeding or some procedures. Repeated exposure can cause the immune system to form antibodies against transfused platelets. These antibodies may make later transfusions less effective. When needed, specialists may select platelets matched for certain immune markers.

Recombinant activated factor VII is a laboratory-made clotting protein. It may be used when platelet transfusions do not work, when relevant antibodies are present, or when suitable platelets are unavailable. Its use requires specialist assessment because it can rarely contribute to unwanted blood clots.

Heavy periods may be managed with antifibrinolytic treatment, hormonal treatment, or both. Iron replacement may be needed if ongoing blood loss has caused iron deficiency.

Planned surgery, dental work, pregnancy, and childbirth require a written bleeding plan. This may include local treatments, medicine, and access to compatible blood products. Emergency and surgical teams should know about the condition before treatment begins.

A stem cell transplant can replace the blood-forming cells that produce abnormal platelets and may cure the condition. However, transplantation carries serious risks. It is generally considered only for selected people with severe bleeding that cannot be controlled adequately by other methods.

Complications

Repeated blood loss can lead to iron deficiency and anaemia. Severe bleeding may require hospital care, particularly when it affects the digestive tract, follows major trauma, or occurs during surgery or childbirth.

Platelet transfusions can trigger antibodies against platelet proteins or human leukocyte antigens, also called HLA. This is known as alloimmunisation. It may cause the body to clear donated platelets quickly, making transfusion less effective.

Heavy menstrual bleeding can disrupt sleep, school, work, and physical activity. It may also cause long-term iron deficiency if blood loss is not recognised and treated.

Pregnancy does not always increase day-to-day bleeding, but miscarriage, delivery, and the weeks after birth can carry a higher bleeding risk. Antibodies against platelet proteins may also affect a newborn's platelet function or count. Specialist planning is important before pregnancy and throughout maternity care.

Prevention

Glanzmann thrombasthenia cannot be prevented because it is inherited. Genetic counselling can explain the chance of passing it on, but it cannot change the genes a person already has.

Some bleeding episodes and treatment complications may be reduced through practical planning:

Aspirin and many non-steroidal anti-inflammatory drugs, such as ibuprofen and naproxen, can further impair platelet function. Some supplements and prescribed medicines can also affect bleeding. Do not stop a prescribed medicine on your own; ask about safer options first.

- Tell medical and dental teams about the condition before procedures
- Carry a medical alert card or emergency treatment plan
- Maintain regular dental care to reduce the need for invasive treatment
- Check medicines with a clinician or pharmacist before taking them
- Use suitable protective equipment for activities with injury risks

Living with it and what to expect

The platelet problem remains throughout life and does not usually become progressively worse on its own. However, bleeding patterns may change with injuries, menstrual periods, pregnancy, dental work, surgery, and other health conditions.

Many people attend school, work, exercise, travel, and have families. Activities may need to be chosen with bleeding risk in mind. Sports involving repeated collisions or a high chance of head injury may need individual discussion with the specialist team.

Regular follow-up usually focuses on bleeding episodes, menstrual symptoms, iron levels, planned procedures, and the response to previous treatments. People who have received platelet transfusions may be tested for antibodies when this could affect future care.

A personal treatment plan should explain what to do for common bleeding, where to seek help, and which treatments have worked before. Keeping this plan available is especially useful

when travelling or attending a medical service unfamiliar with the condition.

Emotional support may also matter. Recurrent bleeding, visible bruises, heavy periods, or uncertainty around procedures can affect confidence and daily plans. A specialist centre or patient organisation may help you connect with counselling, school support, workplace adjustments, or other people living with inherited bleeding conditions.

When to get medical help

Arrange routine follow-up if nosebleeds, gum bleeding, bruising, or menstrual bleeding is becoming more frequent. Tiredness, reduced exercise tolerance, headaches, or pale skin may suggest iron deficiency and should also be assessed.

Contact the haematology team promptly before surgery, dental extraction, pregnancy, or childbirth. Planning is safer than waiting for bleeding to begin.

Seek urgent medical help for:

A head injury needs emergency assessment because bleeding inside the skull may not be visible. Call emergency services for a severe headache after injury, repeated vomiting, confusion, unusual sleepiness, weakness, a seizure, or loss of consciousness.

- Bleeding that is heavy or does not stop with the measures in your plan
- Vomiting blood, coughing up blood, or passing black or bloody stools
- Blood in the urine
- Severe or rapidly increasing menstrual bleeding
- Dizziness, fainting, marked weakness, or difficulty breathing after blood loss
- Significant injury, even if visible bleeding seems limited

Common questions

Is Glanzmann thrombasthenia the same as haemophilia?

No. Haemophilia involves missing or reduced clotting factors, which are proteins dissolved in the blood. Glanzmann thrombasthenia affects how platelets stick together. Both can cause bleeding, but their usual bleeding patterns and treatments are different.

Can I have Glanzmann thrombasthenia with a normal platelet count?

Yes. Most people with the condition have a normal number of platelets. The problem is how those platelets function. This is why a routine blood count may not identify the condition.

Can Glanzmann thrombasthenia be cured?

A stem cell transplant can cure it by replacing the blood-forming cells that make affected platelets. Transplantation has major risks, so it is not routine treatment. Most people are managed with bleeding-control measures rather than transplant.

Can I pass it on to my children?

Yes, but the chance depends on your partner's genes. A person with Glanzmann thrombasthenia usually passes an altered gene copy to every child. Whether a child is affected or is only a carrier depends on whether the other parent also passes on an altered copy.

Can someone with Glanzmann thrombasthenia have periods or become pregnant?

Yes. Periods may be heavy and often need planned treatment. Pregnancy is possible, but delivery and the period after birth can involve significant bleeding. Care should be coordinated by obstetric, anaesthetic, haematology, and blood transfusion teams familiar with the condition.

What painkillers should I avoid?

Aspirin and many anti-inflammatory painkillers like ibuprofen or naproxen can make platelet function worse. Other medicines and supplements may also increase bleeding. A pharmacist or clinician can help identify an appropriate option for the type of pain you have.

Sources

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