CONDITION

Congenital platelet function defects

Congenital platelet function defects are inherited conditions in which platelets do not form blood clots normally.

WHAT IT IS
Inherited disorders that impair platelet clot formation

BODY SYSTEM
Blood and circulation

MAIN SYMPTOMS
Easy bruising, nosebleeds, gum bleeding, heavy periods

WHAT CAUSES IT
Inherited gene changes affecting platelet function

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

MAIN TREATMENT
Bleeding prevention, antifibrinolytics, transfusion, specialist therapies

CAN IT BE CURED?
Sometimes - transplantation cures selected severe inherited defects

IS IT AN EMERGENCY?
Sometimes - heavy or uncontrolled bleeding

LONG-TERM OUTLOOK
Often manageable with specialist planning and prompt bleeding care

Overview

Congenital platelet function defects are a group of inherited conditions that affect how platelets work. Platelets are small blood cells that gather at an injury and help form a plug to stop bleeding.

In these conditions, the platelet count may be normal, but the platelets do not stick, release chemical signals, or join together as they should. Some defects also cause a low platelet count or unusually large platelets.

Examples include Glanzmann thrombasthenia, Bernard-Soulier syndrome, and platelet storage pool disorders. There are also rarer defects involving the receptors, chemical granules, or signalling pathways inside platelets. Some occur alone, while others are part of a condition affecting the immune system, skin colour, hearing, kidneys, or other organs.

The effect can range from mild bruising to serious bleeding after surgery, childbirth, or an injury. Bleeding often comes from moist surfaces such as the nose, mouth, digestive tract, and uterus.

Correct identification matters because routine blood-clotting tests can be normal. Knowing the specific defect helps with planning dental work, surgery, pregnancy, and treatment for bleeding.

Symptoms

Symptoms often begin in childhood, but a mild defect may not be noticed until surgery, dental work, an injury, or the start of menstrual periods.

Possible symptoms include:

Bleeding into joints and muscles is less typical than it is in haemophilia, which affects different clotting proteins. However, it can occur after significant injury or with a severe platelet defect.

Symptoms do not always show how serious a future bleeding event might be. Someone with few day-to-day problems may still need a bleeding plan before an operation. Menstrual bleeding and childbirth can also reveal a condition that previously caused little difficulty.

- Bruising easily or developing large bruises after minor knocks
- Frequent or prolonged nosebleeds
- Bleeding from the gums, especially during dental care
- Heavy or prolonged menstrual bleeding
- Bleeding that lasts longer than expected after cuts
- Excessive bleeding after dental work, surgery, or childbirth
- Small red or purple skin spots caused by bleeding under the skin
- Blood in the urine, vomit, or bowel movements

Causes and risk factors

Congenital platelet function defects are caused by gene changes present from birth. These changes affect proteins that platelets need to attach to damaged blood vessels, release chemical signals, or bind to one another.

Different conditions have different inheritance patterns. Some are autosomal recessive,

meaning a child usually inherits one altered copy of a gene from each parent. The parents are often unaffected carriers. Other defects are autosomal dominant, meaning one altered gene copy may be enough to cause symptoms. Rare types are linked to the X chromosome.

The chance of a recessive condition can be higher when both parents come from the same extended family or a community in which a particular gene change is more common. However, these conditions can occur in any family.

Some inherited syndromes cause a platelet defect along with other features. These may include albinism, repeated infections, hearing loss, kidney problems, or changes in blood cell production.

Medicines and other medical conditions can also impair platelet function, but those problems are acquired rather than congenital. Aspirin and some anti-inflammatory medicines are common examples. A careful review is needed to distinguish an inherited defect from medication effects, von Willebrand disease, liver or kidney problems, and other causes of bleeding.

Diagnosis

Diagnosis usually begins with your personal and family bleeding history. The clinician may ask about nosebleeds, menstrual periods, dental procedures, surgery, childbirth, injuries, and medicines. A standardised bleeding questionnaire may help record this information.

Initial blood tests often include:

PT and aPTT results are often normal in platelet function defects. A normal platelet count also does not rule one out. In Bernard-Soulier syndrome, platelets are usually unusually large and the count may be low.

Specialist platelet tests are needed to investigate how the platelets respond to different chemical triggers. Platelet aggregation testing measures how well they clump together. Secretion testing checks whether they release stored chemicals. Flow cytometry uses labelled antibodies to identify important proteins on the platelet surface.

Some laboratories use electron microscopy to examine the tiny storage granules inside platelets. Genetic testing may identify the altered gene and confirm a specific diagnosis, although it does not find every possible defect.

These tests are technically demanding. Recent illness, a low platelet count, sample transport, and medicines that affect platelets can alter the results. Repeat testing may sometimes be needed at a specialist centre.

- A complete blood count to measure platelets and other blood cells
- A blood film to examine platelet size and appearance
- Tests of clotting time, often called PT and aPTT
- Testing for von Willebrand disease, another inherited bleeding condition

Treatment and management

Treatment is matched to the type of defect, your bleeding history, and the situation. Many people do not need regular treatment. Instead, they have a plan for injuries, procedures, menstrual bleeding, and other times when bleeding risk rises.

For minor bleeding, local measures may be enough. These include firm pressure, wound closure, nasal treatment, or dental products that help a clot stay in place.

Antifibrinolytic medicines help prevent the body from breaking down a newly formed clot too quickly. They are often used for bleeding from the mouth, nose, or uterus and around dental procedures. Treatment must be selected by a clinician because it is not suitable for every type or location of bleeding.

Desmopressin is a medicine that raises certain clot-supporting proteins in the blood. It may help with some mild platelet defects, but its effect is inconsistent and it is not useful for every condition. Specialists may test or assess the likely response before relying on it.

Heavy menstrual bleeding may be managed with antifibrinolytic treatment, hormonal treatment, or both. Iron replacement may be needed if repeated blood loss causes iron deficiency.

Platelet transfusions provide working donor platelets. They may be needed for major bleeding, childbirth, or surgery. Repeated transfusions can cause the immune system to make antibodies against donor platelets, making later transfusions less effective. Specialists may use carefully matched platelets to reduce this risk.

Recombinant activated factor VII, a laboratory-made clotting protein, may be used in selected situations, especially for some people with Glanzmann thrombasthenia who cannot receive effective platelet transfusions. It requires specialist oversight because it can increase the risk of unwanted blood clots.

Stem cell transplantation can replace the blood-forming cells that produce abnormal platelets. It may cure selected severe conditions but carries substantial risks, so it is not routine treatment.

A written bleeding plan or medical alert card can help emergency, dental, and surgical teams

respond appropriately. Tell the relevant team about the condition well before planned procedures. Do not stop prescribed medicine without advice, but check before using aspirin, ibuprofen, naproxen, or other products that can affect platelets.

Complications

Repeated blood loss can cause iron deficiency and anaemia. Anaemia means there are not enough healthy red blood cells to carry oxygen. It may cause tiredness, weakness, headaches, shortness of breath, or pale skin.

Heavy menstrual bleeding can interfere with school, work, exercise, sleep, and social activities. It may also make pregnancy planning more complex.

Major bleeding can occur after surgery, serious injury, or childbirth. Bleeding in the digestive tract or brain is uncommon but can be serious. Some associated genetic syndromes have additional complications unrelated to bleeding.

People who receive platelet transfusions may develop antibodies against donor platelet proteins. This is called alloimmunisation. It can make future transfusions less effective, known as platelet refractoriness.

Living with an unpredictable bleeding risk can also cause worry or lead to avoiding useful activities. A specialist plan can help balance safety with participation in everyday life.

Prevention

The inherited gene change cannot usually be prevented. Nothing a parent did during pregnancy causes a congenital platelet function defect.

Some bleeding episodes can be prevented or limited. Useful steps include:

Regular dental care can reduce the need for more invasive procedures later. Use a soft toothbrush if gum bleeding is a problem, but do not avoid cleaning your teeth unless a dental professional has given different instructions.

Genetic counselling can explain the inheritance pattern and the chance of passing the condition to a child. It can also discuss testing options. This supports informed choices; it does not change the genes of someone who already has the condition.

- Sharing your bleeding plan before surgery, dental work, or childbirth
- Checking medicines and supplements for effects on platelets

- Using protective equipment for activities with a risk of injury
- Treating iron deficiency and heavy menstrual bleeding
- Keeping recommended specialist and dental follow-up appointments
- Wearing medical identification if advised

Living with it and what to expect

The underlying platelet defect is usually lifelong. It does not generally disappear with age, although the pattern of bleeding can change. Childhood nosebleeds may become less prominent, while menstrual bleeding, pregnancy, dental work, or surgery may create new challenges.

A genetic defect does not necessarily become progressively worse. Day-to-day bleeding may remain similar for years. The apparent severity can still change because of medicines, other illnesses, injury, iron deficiency, or procedures.

Many people can attend school, work, travel, exercise, and have families. Activities may need individual planning if they involve collisions, falls, or remote locations where medical help is limited. A haematology team can help assess risk without unnecessarily restricting activity.

Carry information about the exact diagnosis, usual treatment, previous transfusion reactions, and specialist contact details. When travelling, keep essential medical information and prescribed treatment accessible rather than only in checked luggage.

Pregnancy is possible, but planning is important. Bleeding may occur during pregnancy, delivery, or the weeks after birth. The baby may also have inherited the condition. Care is usually coordinated between haematology, obstetric, anaesthetic, and newborn teams.

Routine follow-up may include reviewing bleeding episodes, checking for iron deficiency, updating procedure plans, and discussing new medicines. Follow-up is also a chance to review whether a more precise genetic diagnosis has become available.

When to get medical help

Call emergency services for bleeding that is heavy, rapidly worsening, or not stopping with your usual first-aid measures or bleeding plan.

Emergency assessment is also needed after a significant head injury or for:

Contact your haematology or urgent care team promptly if bleeding lasts longer than usual,

nosebleeds keep returning, bruising suddenly increases, or blood appears in urine or bowel movements. Seek timely advice for menstrual bleeding that is difficult to control or causes dizziness, breathlessness, or marked tiredness.

Routine contact is appropriate when you need to update a bleeding plan, discuss iron symptoms, review medicines, or prepare for dental work, surgery, pregnancy, or travel. Planned procedures should not go ahead without the relevant team knowing about the platelet defect.

- A sudden severe headache, confusion, fainting, seizure, or weakness
- Vomiting blood, coughing up blood, or passing a large amount of blood
- Black, tar-like bowel movements with weakness or dizziness
- Breathing difficulty, chest pain, or signs of shock
- Heavy bleeding during pregnancy or after childbirth

Common questions

Is this the same as having a low platelet count?

No. Platelet function describes how well platelets work, while platelet count describes how many are present. Many congenital platelet function defects occur with a normal count. Some, including Bernard-Soulier syndrome, can affect both number and function.

Will my child inherit it?

The chance depends on the specific gene and its inheritance pattern. In some recessive conditions, children who inherit only one altered gene are unaffected carriers. Dominant and X-linked conditions follow different patterns. Genetic counselling can explain the chance for your family.

Can I take ibuprofen or aspirin?

These medicines can reduce platelet function and may increase bleeding. Do not stop aspirin or another prescribed medicine on your own, particularly if it was prescribed for a heart or circulation condition. Ask your haematology or prescribing team which pain medicines are appropriate for you.

Can I play sports?

Many people can exercise and take part in sports. The safest choice depends on bleeding severity, previous injuries, and how easy it is to obtain care. Activities involving repeated collisions or a high risk of head injury may require extra precautions or an alternative.

Can heavy periods be treated?

Yes. Options can include antifibrinolytic medicines, hormonal treatment, and iron replacement. Treatment can reduce bleeding and protect against iron deficiency. A gynaecology and haematology plan may be helpful when symptoms are difficult to control.

Will the condition get worse as I get older?

The inherited platelet defect usually remains stable rather than steadily worsening. Bleeding problems may seem different as life circumstances change, such as starting periods, becoming pregnant, taking new medicines, or having surgery. New or clearly worsening bleeding should still be assessed.

Sources

- National Heart, Lung, and Blood Institute. Platelet Disorders: Diagnosis and Treatment
- Gomez K, Anderson J, Baker P, Biss T, Jennings I, Lowe G. Clinical and laboratory diagnosis of heritable platelet disorders in adults and children: a British Society for Haematology guideline. British Journal of Haematology. 2021;195:46-72
- Bolton-Maggs PHB, Chalmers EA, Collins PW, et al. A review of inherited platelet disorders with guidelines for their management on behalf of the UKHCDO. British Journal of Haematology. 2006;135:603-633
- Nurden AT. Glanzmann thrombasthenia. Orphanet Journal of Rare Diseases. 2006;1:10
- Berndt MC, Andrews RK. Bernard-Soulier syndrome. Haematologica. 2011;96:355-359