CONDITION

Prothrombin deficiency

Prothrombin deficiency is a rare bleeding condition caused by too little working clotting factor II.

WHAT IT IS
Rare deficiency of clotting factor II

BODY SYSTEM
Blood and circulation

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

WHAT CAUSES IT
Inherited F2 gene changes, rarely acquired

HOW IT IS DIAGNOSED
Clotting tests and a factor II activity assay

MAIN TREATMENT
Clotting factor replacement and antifibrinolytic medicines

CAN IT BE CURED?
No - inherited gene change remains

IS IT AN EMERGENCY?
Sometimes - uncontrolled bleeding or possible brain bleeding

LONG-TERM OUTLOOK
Often manageable with specialist planning and timely treatment

Overview

Prothrombin deficiency is a rare bleeding condition. It is also called factor II deficiency because prothrombin is clotting factor II, one of the proteins that helps blood form a stable clot.

Prothrombin is made in the liver. When bleeding starts, it is changed into thrombin. Thrombin then helps create strands of fibrin, a protein that strengthens the blood clot. If too little prothrombin is present or it does not work properly, bleeding can continue longer than expected.

Inherited prothrombin deficiency is different from prothrombin thrombophilia , which is linked to an increased risk of blood clots. Although both involve the F2 gene, they have different effects and need different management.

There are two main forms of inherited deficiency. In hypoprothrombinemia , the body makes too little prothrombin. In dysprothrombinemia , the body may make a near-normal amount, but the protein does not work correctly.

The effect can range from mild bleeding noticed only after an operation to serious spontaneous bleeding. Your factor II level is important, but it does not always predict every bleeding event. Your personal and family bleeding history also helps guide care.

Symptoms

Symptoms may first appear in infancy or childhood, but mild deficiency can remain unnoticed until dental work, an operation, childbirth, or an injury.

Possible symptoms include:

In a newborn, prolonged bleeding from the umbilical cord or after a medical procedure can be an early sign. Severe deficiency can rarely cause bleeding in or around the brain.

Joint bleeding may cause pain, warmth, swelling, or reduced movement. Repeated bleeding into the same joint can eventually damage it. Muscle bleeding can cause swelling, pain, weakness, tingling, or pressure on nearby nerves.

Some people with a mild deficiency have no day-to-day symptoms. Others mainly notice heavy periods, frequent nosebleeds, or unusually prolonged bleeding after procedures.

- Easy or unexplained bruising
- Frequent or prolonged nosebleeds
- Bleeding from the gums or mouth
- Bleeding that continues after a cut, injection, dental procedure, or operation
- Heavy or prolonged menstrual periods
- Bleeding after childbirth
- Blood in urine or stool
- Bleeding into muscles or joints
- Internal bleeding after an injury

Causes and risk factors

Inherited prothrombin deficiency is caused by changes in the F2 gene. This gene gives the

body instructions for making prothrombin.

It is usually inherited in an autosomal recessive pattern. This means a person generally inherits an altered copy of the gene from each parent. Parents who each carry one altered copy often have no symptoms, although some carriers may have a mildly reduced factor level or a bleeding tendency.

The inherited condition affects people of any sex. It may be more likely when both biological parents come from the same extended family or from a population in which a particular F2 gene change is more common.

Low factor II can also be acquired rather than inherited. Possible causes include severe liver problems, vitamin K deficiency, medicines that reduce vitamin K-dependent clotting factors, and conditions that consume clotting factors throughout the bloodstream. Rarely, an immune-related condition can cause the body to remove prothrombin from the blood.

Acquired low prothrombin is not passed from parent to child. Its treatment and outlook are based on the underlying cause.

Diagnosis

Diagnosis begins with your bleeding history, family history, medicines, and any liver, nutrition, or immune problems. A physical examination may look for bruising, joint swelling, anemia, or signs of an underlying condition.

Initial blood tests usually include prothrombin time , or PT, and activated partial thromboplastin time , or aPTT. These tests measure how long blood takes to clot through different parts of the clotting process. Both are often prolonged in significant factor II deficiency, but these results do not identify the exact missing factor.

A factor II activity assay is used to confirm that prothrombin activity is reduced. Additional tests may include:

Genetic testing can support the diagnosis and help with family counselling, but it may not be necessary in every case. A normal family history does not rule out the condition.

Testing should also distinguish factor II deficiency from the prothrombin G20210A variant. That variant is usually associated with increased clotting risk, not a lack of working prothrombin.

- Assays for other clotting factors
- A mixing study to check for an inhibitor interfering with clotting

- A prothrombin antigen test to measure how much protein is present
- Liver tests and tests for vitamin K-related causes
- Genetic testing for changes in the F2 gene

Treatment and management

Treatment is planned around the severity of bleeding, your previous bleeding pattern, factor II activity, and whether you are having surgery or giving birth. Care is often coordinated through a specialist bleeding disorders centre.

Small mouth or nose bleeds may be treated with local pressure and an antifibrinolytic medicine . These medicines help prevent a newly formed clot from breaking down too quickly. They may also be used for dental work or heavy menstrual bleeding when medically appropriate.

More significant bleeding may require replacement of the missing clotting factor. There is no widely available concentrate containing only factor II. Replacement may therefore use:

Prothrombin complex concentrate can raise factor levels without the volume of plasma, but it may increase the risk of an unwanted blood clot. Fresh frozen plasma requires a larger fluid volume and can cause transfusion reactions. The choice requires specialist assessment.

People with heavy menstrual bleeding may be offered medicines that reduce blood loss. Iron testing and iron replacement may be needed if ongoing bleeding causes iron-deficiency anemia.

Regular preventive replacement is not required for everyone. It may be considered for selected people with severe deficiency or repeated serious bleeding. A written plan is important before surgery, dental procedures, pregnancy, childbirth, or invasive tests.

If low factor II is acquired, treatment focuses on the cause. This may include correcting vitamin K deficiency, managing liver or immune problems, or reviewing medicines that affect clotting.

- Prothrombin complex concentrate , which contains factor II and several other clotting factors
- Fresh frozen plasma , the liquid portion of donated blood containing clotting proteins

Complications

Blood loss over time can cause iron-deficiency anemia. This may lead to tiredness, weakness, headaches, shortness of breath with activity, or difficulty concentrating.

Repeated bleeding into joints can cause long-term pain, stiffness, and loss of movement. A large muscle bleed may press on nerves or blood vessels. Internal bleeding can damage an organ if it is not recognised and treated.

Bleeding in or around the brain is uncommon but serious. It can occur after a head injury or, in severe deficiency, without an obvious injury.

Treatment also has possible complications. Blood products can cause allergic or transfusion reactions. Prothrombin complex concentrate may contribute to an unwanted clot, particularly when other clotting risks are present. Specialist monitoring helps balance bleeding control against these risks.

Prevention

Inherited prothrombin deficiency cannot currently be prevented because it is caused by a gene change present from birth. Genetic counselling can explain inheritance and testing options, but it does not remove the condition.

Bleeding complications can sometimes be reduced through planning. Tell your bleeding disorders team before operations, dental work, pregnancy, or childbirth. Carry a bleeding-disorder card or medical alert information, and make sure relevant clinicians know your diagnosis.

Check before taking medicines or supplements that may affect bleeding. Do not stop a prescribed medicine without medical advice. Protective equipment for activities with a risk of head injury may also be appropriate.

Acquired low factor II may sometimes be preventable or reversible when it results from a medicine, poor vitamin K absorption, or another treatable cause. Prevention is based on addressing that specific cause rather than on general lifestyle changes.

Living with it and what to expect

Inherited prothrombin deficiency is lifelong. The underlying gene change does not go away, although the pattern of bleeding can change with age, injuries, menstruation, pregnancy, procedures, and other health conditions.

Many people with mild deficiency have few restrictions and need treatment mainly before procedures or after significant injuries. People with more severe deficiency may need closer follow-up, faster access to treatment, and occasionally preventive replacement.

A personal bleeding plan should explain what to do after an injury, where treatment is available, and which blood product or medicine has been recommended. Keep this plan accessible when travelling, starting school or work, or changing healthcare services.

Children should be encouraged to remain active while avoiding activities with an unusually high risk of serious head or abdominal injury. An individual plan can help balance safety with normal development and participation.

Pregnancy and childbirth require early planning with obstetric, anesthesia, and bleeding-disorder teams. Factor levels and bleeding history help determine what support may be needed during delivery and afterward. Heavy bleeding after birth can occur even if the pregnancy itself was uncomplicated.

Regular follow-up may include review of bleeding episodes, joint symptoms, menstrual blood loss, iron levels, and upcoming procedures. Stable symptoms do not usually require emergency assessment, but new or increasing bleeding should be discussed with your care team.

When to get medical help

Seek emergency care for:

After a significant head, neck, abdominal, or back injury, contact emergency services or your specialist treatment service even if there is no visible bleeding. Internal bleeding may not be obvious at first.

Contact your bleeding disorders team promptly for a painful or swollen joint, blood in urine, a large unexplained bruise, persistent nosebleeds, or menstrual bleeding that is much heavier than usual.

Routine follow-up is appropriate for reviewing mild recurring symptoms, iron deficiency, activity choices, family testing, or plans for future dental work, surgery, pregnancy, or travel.

- A head injury followed by severe headache, vomiting, confusion, unusual sleepiness, weakness, seizure, or loss of consciousness
- Bleeding that is heavy, rapidly increasing, or will not stop with firm pressure
- Vomiting blood, coughing up blood, or passing black or heavily blood-stained stool
- Sudden severe abdominal, back, chest, or pelvic pain
- Rapidly increasing muscle swelling, numbness, weakness, or severe pain
- Heavy bleeding with faintness, breathing difficulty, marked weakness, or pale, clammy skin

Common questions

Is prothrombin deficiency the same as hemophilia?

No. Both are inherited bleeding conditions, but classic hemophilia involves factor VIII or factor IX. Prothrombin deficiency involves factor II and is generally managed as a rare clotting factor deficiency.

Is it the same as the prothrombin gene mutation that causes blood clots?

No. The prothrombin G20210A variant can increase the risk of unwanted blood clots. Inherited prothrombin deficiency causes too little working factor II and usually increases bleeding risk.

Can I have prothrombin deficiency without symptoms?

Yes. Mild deficiency may cause no obvious daily symptoms. It may first be found after prolonged bleeding from an operation, dental work, childbirth, or an injury.

Can prothrombin deficiency be cured?

The inherited condition cannot currently be cured because the underlying gene change remains. Treatment can replace clotting factors, control bleeding, and reduce risks around procedures. Acquired low factor II may improve if its cause can be corrected.

Can I exercise or play sports?

Many people can remain active. The safest activities depend on your bleeding history and severity. Activities with a high risk of head injury or major collision may require extra precautions or may not be suitable for someone with severe deficiency.

Will my children inherit it?

That depends on which F2 gene changes you and the other biological parent carry. Because the condition is usually autosomal recessive, a genetic counsellor can explain the chance of a child being affected or being a carrier.

Sources

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