CONDITION

Hemophilia B

Hemophilia B is an inherited condition that reduces blood clotting because factor IX is missing or too low.

WHAT IT IS
Inherited deficiency of blood-clotting factor IX

BODY SYSTEM
Blood and circulation

MAIN SYMPTOMS
Prolonged bleeding, easy bruising, joint bleeding, muscle bleeding

WHAT CAUSES IT
Inherited change in the F9 gene

HOW IT IS DIAGNOSED
Clotting tests and factor IX activity testing

MAIN TREATMENT
Factor replacement and other bleeding-prevention therapies

CAN IT BE CURED?
No - current treatments control bleeding

IS IT AN EMERGENCY?
Sometimes - head injury or uncontrolled bleeding

LONG-TERM OUTLOOK
Usually active lives with ongoing specialist care

Overview

Hemophilia B is a lifelong condition in which the blood does not clot normally. It happens when the body has too little working clotting factor IX. Clotting factors are proteins that work together to form a stable blood clot after an injury.

The condition is sometimes called factor IX deficiency. Unlike a common cut that simply takes a little longer to stop bleeding, hemophilia can cause bleeding deep inside joints, muscles, or other parts of the body. Some bleeding starts without a clear injury.

Hemophilia B can be mild, moderate, or severe. This is based mainly on the amount of factor IX activity in the blood. People with severe hemophilia are more likely to have spontaneous bleeding. People with mild hemophilia may not notice a problem until surgery, dental work, childbirth, or a significant injury.

Hemophilia B most often affects males because of the way it is inherited. Females can carry the genetic change and may also have low factor IX levels or bleeding symptoms. Anyone with symptoms deserves appropriate testing rather than being described only as a "carrier."

With specialist care and effective treatment, many people with hemophilia B attend school, work, exercise, travel, and have families.

Symptoms

Symptoms depend on the factor IX level, treatment, injuries, and other health factors. They may begin in infancy or appear later.

Possible symptoms include:

A joint bleed may cause tingling, warmth, stiffness, pain, or swelling. Young children may avoid using an arm or leg or may begin limping. Repeated joint bleeding can damage cartilage and lead to long-term pain and reduced movement.

Bleeding into a large muscle can press on nerves or blood vessels. This may cause numbness, increasing pain, weakness, or a pale or cool limb.

Bleeding in the brain is uncommon but serious. It can happen after a head injury or sometimes without an obvious injury, particularly in severe hemophilia.

- Bleeding that lasts longer than expected after cuts, surgery, or dental work
- Large or frequent bruises
- Nose or mouth bleeding that is difficult to stop
- Bleeding into joints, especially the ankles, knees, or elbows
- Bleeding into muscles, causing pain, swelling, tightness, or weakness
- Blood in urine or stool
- Heavy menstrual bleeding
- Excessive bleeding after childbirth

Causes and risk factors

Hemophilia B is caused by a harmful change in the F9 gene, which provides instructions for making factor IX. The gene is on the X chromosome.

A male usually has one X chromosome. If that chromosome carries the F9 change, he will have

hemophilia B. A female usually has two X chromosomes. If one carries the change, her other copy may provide enough factor IX, but this is not guaranteed. Some females have factor levels low enough to cause mild, moderate, or, rarely, severe bleeding.

The genetic change may be passed through a family. It can also arise for the first time in a child, so hemophilia B is possible without a known family history.

The chance of inheriting the condition is determined by which parent carries the genetic change and which chromosomes a child inherits. Genetic counselling can explain this in the context of an individual family without assuming anyone's sex, gender identity, or plans for pregnancy.

Hemophilia B is not caused by diet, exercise, or anything a parent did during pregnancy. It is not contagious.

Rarely, a person can develop an antibody that interferes with factor IX without having inherited hemophilia. This is a different problem called acquired factor IX deficiency and requires separate assessment.

Diagnosis

Diagnosis begins with a medical and family history. A clinician may ask about bruising, joint or muscle bleeding, bleeding after procedures, menstrual bleeding, and relatives with similar problems.

Initial blood tests often include:

A factor IX activity test confirms that factor IX is reduced and helps classify the condition as mild, moderate, or severe. Other testing may be needed because liver disease, vitamin K deficiency, medicines, or antibodies against clotting factors can also affect clotting results.

Genetic testing can identify the F9 gene change. It may help confirm inheritance, test relatives, or support pregnancy-related planning. A genetic result is useful but is not always required to diagnose hemophilia B.

Babies with a family history may be tested soon after birth. Factor IX levels are naturally lower in newborns, especially those born early, so mild hemophilia may require repeat testing later.

- Activated partial thromboplastin time, or aPTT: measures part of the clotting process and may be prolonged
- Prothrombin time, or PT: often remains normal in hemophilia B
- Complete blood count: checks blood cells and signs of blood loss

Treatment and management

Hemophilia treatment is usually coordinated through a specialist hemophilia treatment centre. The plan is based on bleeding history, factor IX level, age, joint health, lifestyle, other conditions, and whether antibodies to treatment are present.

The main treatment is factor IX replacement. This replaces the missing clotting protein through an infusion into a vein. Standard and extended half-life products are available. Extended half-life products remain active longer.

Factor may be given in two main ways:

People with severe hemophilia commonly use prophylaxis. Some people with milder disease need treatment mainly around surgery, dental procedures, injuries, or bleeding episodes. The exact product and schedule must be planned individually.

Non-factor medicines can prevent bleeding without directly replacing factor IX. Whether one is suitable depends partly on age and whether the person has an inhibitor. An inhibitor is an antibody that blocks factor IX treatment. Bypassing medicines may be used when an inhibitor prevents replacement factor from working.

Gene therapy is available for some adults with hemophilia B. It delivers a working factor IX gene to liver cells, with the aim of increasing factor production. It may greatly reduce bleeding and the need for prophylaxis, but the response is not identical for everyone and may change over time. It is not considered a guaranteed cure. Careful assessment and long-term monitoring are required.

Physical therapy can help preserve joint movement, muscle strength, and balance. Dental care is also important because preventing gum disease and tooth decay reduces the need for invasive treatment.

Surgery, dental procedures, and childbirth need advance planning with the hemophilia team. Do not stop or start medicines because of hemophilia without discussing them with the relevant care team.

- Prophylaxis: regular treatment intended to prevent bleeding
- On-demand treatment: treatment given when a bleed occurs

Complications

Repeated joint bleeding can cause hemophilic arthropathy, meaning lasting joint damage from blood and inflammation inside the joint. This may lead to chronic pain, stiffness, weakness, and

difficulty moving.

Deep muscle bleeding can damage nerves or limit blood flow. Internal bleeding may also cause anemia, which means too few healthy red blood cells.

Some people develop inhibitors against factor IX. These antibodies make usual factor replacement less effective. Inhibitors are less common in hemophilia B than in hemophilia A, but they can be difficult to manage. Some people with factor IX inhibitors can also have serious allergic reactions to factor treatment.

Modern factor products have strong safeguards against infection. People who received older blood-derived products decades ago may have acquired hepatitis or HIV and may need continuing follow-up for those infections.

Living with a lifelong bleeding condition can also affect mood, relationships, school, work, or confidence with physical activity. Emotional and practical support can be part of comprehensive hemophilia care.

Prevention

Hemophilia B itself cannot currently be prevented because it is caused by an inherited or newly occurring genetic change.

Many bleeding episodes and complications can be prevented or reduced. Helpful measures include following the agreed preventive treatment plan, recognizing bleeds early, and attending routine hemophilia and joint assessments.

Regular, suitable physical activity strengthens muscles and protects joints. Lower-impact activities are often easier to manage, but the safest choice depends on treatment, joint health, and the risk of injury.

Avoid aspirin and nonsteroidal anti-inflammatory drugs such as ibuprofen unless the hemophilia team says they are appropriate. These medicines can interfere with platelets, the blood cells that help form clots. Check before using supplements or other medicines that may affect bleeding.

Good dental hygiene, seat belts, helmets for relevant activities, and protective equipment can reduce avoidable injuries. Vaccinations remain important, but the injection method may need planning.

Carrying a medical identification card or wearing medical alert jewellery can help others provide appropriate care in an emergency.

Living with it and what to expect

Hemophilia B remains present throughout life. Factor IX levels caused by the inherited condition generally do not return to normal on their own. However, bleeding patterns can change with preventive treatment, physical activity, joint health, ageing, and other medical conditions.

Many children treated early with effective prophylaxis grow up with much less joint damage than earlier generations experienced. They can usually attend school and take part in a range of activities. Treatment may need adjustment as body size, routines, and activity levels change.

Adults may need continuing attention to joints that were damaged by earlier bleeding. Physical therapy, pain management, preventive treatment, and sometimes orthopedic care can help preserve mobility.

Travel and work are usually possible with planning. This may include carrying enough treatment, a medical letter, emergency contact details, and information about nearby hemophilia services.

Routine follow-up checks bleeding frequency, treatment effectiveness, factor levels when relevant, inhibitor development, joint health, and quality of life. People who receive gene therapy need long-term monitoring, including blood tests for liver health and treatment response.

Pregnancy and birth require specialist planning for someone with low factor IX or an F9 gene change. Factor IX does not reliably rise during pregnancy, so bleeding risk must be assessed before delivery and afterward. A baby who may have hemophilia may also need a planned approach to delivery and newborn testing.

When to get medical help

Seek emergency care after any significant head injury or if there may be bleeding in the brain. Warning signs include a sudden or severe headache, repeated vomiting, unusual sleepiness, confusion, seizure, fainting, weakness, poor balance, or changes in vision or speech. Symptoms can be delayed.

Emergency assessment is also needed for:

If you have an emergency treatment plan, follow it while arranging care. Tell emergency staff about hemophilia B promptly. Treatment for a suspected serious bleed may need to begin before scan results are available.

Contact the hemophilia team promptly for a possible joint or muscle bleed, repeated nose or

mouth bleeding, blood in urine, unusually heavy menstrual bleeding, or bleeding that does not respond as expected to the usual plan.

Routine appointments are appropriate for reviewing preventive treatment, new bruising patterns, ongoing joint discomfort, planned dental work, surgery, pregnancy planning, or concerns about medicines.

- Difficulty breathing or swallowing
- Bleeding or swelling in the throat or neck
- Uncontrolled external bleeding
- Severe abdominal, back, hip, or groin pain
- A painful swollen muscle with numbness, weakness, or a cool limb
- Major trauma, such as a serious fall or vehicle collision
- Vomiting blood or passing a large amount of blood in stool

Common questions

Is hemophilia B the same as hemophilia A?

No. Hemophilia A involves factor VIII, while hemophilia B involves factor IX. Their bleeding symptoms can be similar, but the replacement products and some treatment options differ.

Can females have hemophilia B?

Yes. Females with an F9 gene change may have normal, reduced, or very low factor IX levels. They can experience heavy periods, bleeding after procedures or childbirth, and other hemophilia symptoms.

Can someone with hemophilia B play sports?

Often, yes. Regular activity can strengthen muscles and support joint health. The best choices depend on bleeding control, joint condition, treatment, and injury risk. Higher-contact activities require individual discussion and planning.

Will my child outgrow hemophilia B?

No. Inherited factor IX deficiency is lifelong. Treatment can prevent many bleeds and allow an active life, but it does not make the underlying genetic change disappear.

Is gene therapy a cure for hemophilia B?

Gene therapy can raise factor IX levels and reduce bleeding for some eligible adults. It does not work equally well for everyone, requires long-term monitoring, and is not currently described

as a certain permanent cure.

Can I take ibuprofen if I have hemophilia B?

Ibuprofen can increase bleeding by affecting platelet function. Do not use it unless a clinician familiar with your hemophilia has said it is suitable. Other pain-relief options may be safer for you.

Sources

- World Federation of Hemophilia: Guidelines for the Management of Hemophilia, third edition , Haemophilia , 2020
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- GeneReviews: Hemophilia A and Hemophilia B , University of Washington and National Center for Biotechnology Information
- US Food and Drug Administration: HEMGENIX prescribing information
- National Bleeding Disorders Foundation: MASAC recommendations concerning products licensed for hemophilia treatment