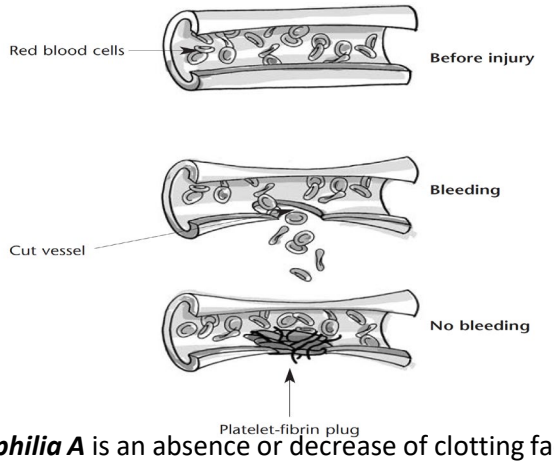
boston hemophilia center

Patient & Family Education Sheet
Hemophilia

What is Hemophilia? Hemophilia is an inherited bleeding disorder - the blood does not clot normally because of a missing or poorly functioning clotting factor protein. The severity of hemophilia may be mild, moderate, or severe depending on the level of clotting factor in the blood. Patients with hemophilia are optimally care for by the integrated care model known as a Hemophilia Treatment Center (HTC). Our primary HTC team includes a physician, nurse practitioner, nurse, social worker, physical therapist and data coordinator.

The Basics

Coagulation: Clotting proteins, also called ***clotting factors***, work with other blood components – such as platelets and von Willebrand factor – to form a blood clot.

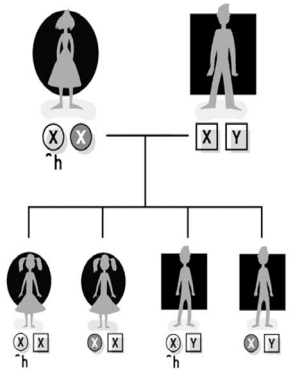


Hemophilia A is an absence or decrease of clotting factor VIII (8); ***Hemophilia B*** is an absence or decrease of factor IX (9). When either of these factors is absent or low, easier and more frequent bleeding can result.

Factor levels:
Normal levels of factor VIII and IX = 50-150%
Mild hemophilia =5-<50%
Moderate hemophilia = 1-<5%
Severe hemophilia = <1%

The Cause

How does one get Hemophilia? Hemophilia A and B are both genetic disorders. The genes that control the production of factor VIII and IX are on the X-chromosome. When a mutation (variant) arises in either of these genes it can prevent the body from making the normal clotting factor. About 1/3 of cases develop from new mutations with no family history. This abnormal gene can be passed from parent to child on the X-chromosome.



A father will pass his "hemophilia" gene to all of his daughters; they will be **hemophilia carriers**. Some hemophilia carriers also have low factor VIII or IX levels and also have hemophilia. A mother with a "hemophilia" gene can pass the gene to her sons or daughters. If this happens her son will have hemophilia and her daughter will be a hemophilia carrier and could have hemophilia if her factor VIII levels are low.

Signs & Symptoms of Bleeding

Individuals with mild or moderate hemophilia usually only have bleeding symptoms with trauma/injury or procedure. Patients with severe hemophilia and some patients with moderate hemophilia can bleed without any clear cause (spontaneous).

People with hemophilia often bruise easily. This is common and usually not serious. It can typically occur in places like the chest (at pressure points where parents hold young children), arms and also on the knees and lower legs.

Symptoms of muscle and joint bleeds include pain, swelling, loss of ability to move (the arm or leg) normally. Often there is no bruising to indicate the bleed, so recognizing these symptoms is important. Changes in level of alertness, vomiting, unusual crying should prompt a call to your hemophilia team.

Who do I call if I am worried about a bleed?
Call your hemophilia team (617) 355-6101 or page the hematologist after hours (617) 355-6363. We will review the symptoms with you and arrange for in-person evaluation if necessary.

childrenshospital.org/BHC

Achieving wellness for individuals with bleeding disorders
Pediatric Bleeding Disorders Program • (617) 355-6101

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Treatment

Can Hemophilia be cured?

Not yet, but there are many treatments to improve factor levels and decrease bleed risk. Early gene therapy treatments that typically improve levels to at least the mild hemophilia range are now becoming available.

How is Hemophilia treated?

Bleeding into the joints or muscles is treated by intravenous (IV) infusion of specific clotting factor concentrate. In some cases, this requires coming to the hospital (especially early on); however, patients can also be treated at home.

Home infusion nurses help with these infusions at first, then parents and patient learn how to do the infusion.

How often is Hemophilia treatment needed?

This depends on the individual patient and type of hemophilia. Infants and those with mild disease are usually treated on-demand (when a bleed occurs). Patients with severe hemophilia begin **prophylaxis** (bleed prevention treatment) within the first 1-2 years of life. The prophylaxis medication, dose and schedule of administration depends on whether you have hemophilia A or B.

Hemophilia A (Factor VIII Deficiency)

- Many treatment products are available to prevent and manage bleeding symptoms, including both factor concentrates and non-factor (emicizumab).
- For mild hemophilia, a nasal spray (high-dose DDAVP/desmopressin) may be an option.

Hemophilia B (Factor IX Deficiency)

- Several factor products are available, both short acting and longer acting.
- Children may infuse twice per week or once weekly depending on the factor product.
- There are no alternate replacement options.

Common Questions

How common is hemophilia?

Approximately 1 in 4,000 males born in the U.S. has hemophilia. Races and economic groups are affected equally.

Is there anything I need to know about Immunizations?

Individuals with hemophilia should follow the routine schedule for immunizations; however, immunizations should be administered subcutaneously (SQ) if possible and with application of ice to reduce the risk of a muscle bleed.

Is it okay to bring my child to the Dentist?

Yes! We strongly encourage building a relationship with a dentist early. Routine dental care can prevent cavities and gingivitis and decrease the need for dental procedures in the future. While routine cleanings do not require specific hemophilia treatment, dental procedures should be discussed in advance with your hemophilia team to ensure a bleed prevention plan is in place.

What should I tell Daycare or School staff?

Adults that participate in the care of your child should be made aware that your child has hemophilia and given instructions on what to do if there is a concern. We offer hemophilia education to daycares and schools to provide a safety conscience environment for your child without being unnecessarily restrictive. We encourage individuals with hemophilia to have normal participation in school, work, physical activities and social events.

Is it okay to travel?

Yes, it is fine for patients with hemophilia to travel. It is important to travel with the supplies necessary to treat a bleed. We will provide you with a travel letter to help guide physicians. Our hematology team is available to you 24/7 by paging the hematologist on-call (617) 355-6363.

How long do people with Hemophilia live?

With modern treatment children born with hemophilia can expect to live long, full lives.

Helpful Websites & Educational Resources

Boston Hemophilia Center www.childrenshospital.org/centers-and-services/hemophilia-program
National Bleeding Disorders Foundation www.hemophilia.org
New England Hemophilia Association www.newenglandhemophilia.org
World Foundation of Hemophilia www.wfh.org
HemAware www.hemaware.org
LA Kelley Communications www.kelleycom.com

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