Hemophilia A: Disease Overview

General Information

Hemophilia A is the most common type of hemophilia. It is also known as factor VIII deficiency or classic hemophilia. It is largely an inherited disorder in which one of the proteins needed to form blood clots is missing or reduced. In about 30% of cases, there is no family history of the disorder and the condition is the result of a spontaneous gene mutation.

Approximately one in 5,000 males born in the United States has hemophilia. All races and economic groups are affected equally.

When a person with hemophilia is injured, he does not bleed harder or faster than a person without hemophilia, he bleeds longer. Small cuts or surface bruises are usually not a problem, but more traumatic injuries may result in serious problems and potential disability (called "bleeding episodes").

Mild, Moderate, and Severe Hemophilia

• People with mild hemophilia usually have problems with bleeding only after serious injury, trauma, or surgery. In many cases, mild hemophilia is not discovered until an injury or surgery or tooth extraction results in unusual bleeding. The first episode may not occur until adulthood.

• People with moderate hemophilia, about 15% of the hemophilia population, tend to have bleeding episodes after injuries. They may also experience occasional bleeding episodes without obvious cause. These are called "spontaneous bleeding episodes."

• People with severe hemophilia, about 60% of the hemophilia population, have bleeding following an injury and may have frequent spontaneous bleeding episodes, often into the joints and muscles.

Symptoms

- Bleeding into joints, with associated pain and swelling
- Blood in the urine or stool
- Bruising
- Gastrointestinal tract and urinary tract hemorrhage
- Prolonged bleeding from cuts, tooth extraction, and surgery
- Spontaneous bleeding

Treatment

Standard treatment involves replacing the missing clotting factor. The amount of factor VIII concentrates needed depends on how severe the bleeding is, the site of the bleeding, and the size of the patient.

Mild hemophilia may be treated with desmopressin (DDAVP), which helps the body release factor VIII that is stored within the lining of blood vessels.

To prevent a bleeding crisis, people with hemophilia and their families can be taught to give factor VIII concentrates at home at the first signs of bleeding. People with severe forms of the disease may need regular preventative treatment.

Depending on the severity of the disease, DDAVP or factor VIII concentrate may be given before having dental extractions and surgery to prevent bleeding.

Immunization with Hepatitis B vaccine is necessary because of the increased risk of exposure to hepatitis due to frequent blood infusions.

Patients who develop an inhibitor to factor VIII may require treatment with other clotting factors such as factor VIIa, which can help with clotting even without any factor VIII.

Outlook (Prognosis)

With treatment, the outlook is good. Most people with hemophilia are able to lead relatively normal lives. A small percentage of people with hemophilia will develop inhibitors to factor VIII, and may die from loss of blood.

People with hemophilia A should establish regular care with a hematologist (blood doctor), especially a doctor who is associated with a hemophilia treatment center. The ability to have quick and easy access to medical records documenting the level of factor VIII that the person has had, the history of factor transfusions (including the types and amounts), any complications, and the type and amount of any inhibitors can be lifesaving in the event the person with hemophilia is in an emergency situation.

Possible Complications

Inhibitors: In some patients with hemophilia, the immune system produces an antibody that inhibits the action of replacement blood products and prevents clot formation. This antibody is known as an inhibitor. The presence of an inhibitor makes the treatment of bleeding episodes more difficult. An inhibitor destroys the clotting factor before it has a chance to stop the bleeding. The reason inhibitors develop is uncertain; however, they occur more frequently in people with severe forms of hemophilia, particularly factor VIII deficiency, because of their need for more frequent infusions. Inhibitors tend to develop within the first one to three years of treatment, typically between the 50^{th} and 100^{th} exposure days.

Joint Damage: One of the major complications of hemophilia is joint damage or "hemophilic arthropathy" that can occur when there is bleeding into joints. This is the most common clinical complication of hemophilia. Bleeding into knees, elbows, ankles, shoulders, and hips can lead to chronic swelling and later joint deformity. Many people with severe hemophilia can suffer from painful, debilitating joint bleeds and associated mobility issues that severely impede their quality of life.

HIV/AIDS: In the late 1970s and 80s people with hemophilia were treated with blood products derived from thousands of donors. When the U.S. blood supply became contaminated by HIV, the products used as treatment for thousands of people with bleeding disorders also became contaminated. More than 50% of the hemophilia population became infected with HIV prior to 1985. However, HIV transmission by factor concentrates in the United States has not occurred since 1986 due to viral inactivation methods used in manufacturing blood products.

Hepatitis: There are six main hepatitis viruses which cause problems ranging from mild chronic infections to liver failure. Almost 95% of all hepatitis cases are hepatitis A, B, or C. Some hepatitis viruses can be asymptomatic for many years and may never become chronic. Others can progress to liver cancers, end stage liver disease, and other life threatening conditions. Symptoms may include fatigue, nausea, vomiting, joint aches, liver tenderness and enlargment, and weight loss. As a result of newer treatment processes for clotting factor, there have been no reports of hepatitis C transmission from clotting factor since 1997. Transmission of hepatitis A, however, remains a risk for people with bleeding disorders who use plasma-derived products. This is because hepatits A virus can resist the viral inactivation methods used to manufacture plasma products. Vaccination against hepatits A and hepatitis B is currently available and is recommended for all patients with bleeding disorders.

** For further information or questions, please feel free to contact Family Factor at 251-633-8090 or Toll Free at 1-877-611-0004.*

REFERENCES

1. National Hemophilia Foundation, "Hemophilia A (Factor VIII Deficiency)". National Hemophilia Foundation. 05/06/2009 <http://www.hemophilia.org/NHFWeb/MainPgs/MainNHF.aspx?menuid=180&contentid=45&rptname=bleeding>.
2. Medical Encyclopedia, "Hemophilia A". Medline Plus Medical Encyclopedia. 05/06/2009 <http://www.nlm.nih.gov/medlineplus/ency/article/000538.htm>.