Hemophilia B: Disease Overview

General Information

Hemophilia B is the second most common type of hemophilia. It can also be known as factor IX deficiency, or Christmas disease. It was originally named “Christmas disease” for the first person diagnosed with the disorder back in 1952.

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.

Hemophilia B is far less common than Hemophilia A. Occurring in about one in 25,000 male births, hemophilia B affects about 3,300 individuals in the United States. 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 and associated pain and swelling
- Blood in the urine or stool
- Bruising
- Excessive bleeding following circumcision
- Gastrointestinal tract and urinary tract hemorrhage
- Nosebleeds
- Prolonged bleeding from cuts, tooth extraction, and surgery
- Spontaneous bleeding

Treatment

Standard treatment is infusion of factor IX concentrates to replace the defective clotting factor. The amount infused depends upon the severity of bleeding, the site of the bleeding, and the size of the patient.

To prevent a bleeding crisis, people with hemophilia and their families can be taught to administer factor IX concentrates at home at the first signs of bleeding. People with severe forms of the disease may need ongoing, preventive infusions.

Depending on the severity of the disease, factor IX concentrate may be given prior to dental extractions and surgery to prevent bleeding.

Outlook (Prognosis)

The outcome is usually good with treatment. Most people with hemophilia are able to lead relatively normal lives. A small percentage of people develop inhibitors of factor IX, and may die from loss of blood.

People with hemophilia B should establish an ongoing relationship with a hematologist, especially one associated with a hemophilia treatment center. The ability to have quick and easy access to medical records describing their level of factor IX, history of transfusions (including the type and amount), any complications they've had, 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 50th and 100th 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

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