

# Characteristics of Diagnosis and Treatment for Hemophilia

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## DESCRIPTION

A genetic bleeding ailment called hemophilia causes the blood to clot improperly. Low amounts of either factor VIII or factor IX are seen in hemophiliacs. The quantity of factor in a person's blood determines the severity of their hemophilia. As the amount of the component declines, there is a greater chance of bleeding, which can have serious health consequences.

The majority of instances include middle-aged or elderly adults, young women who have recently given birth or are in the late stages of pregnancy, or older people. The correct medical care frequently accelerates the recovery of this ailment.

#### Causes

Hemophilia is caused by a mutation or variation in one of the genes those codes for the clotting factor proteins necessary to form a blood clot. The clotting protein may no longer function at all or not at all as a result of this modification or mutation. The X chromosome contains these genes. Males receive the X and Y chromosomes from their mothers and fathers, respectively.

Therefore, if a guy inherits an X chromosome that has a mutation in either the factor VIII or factor IX gene, they may develop a disease similar to hemophilia. Hemophilia can also affect women however this is far less common. These conditions either result in the involvement of both X chromosomes or in the involvement of one X chromosome while the other is missing or inactive. These ladies may experience bleeding symptoms comparable to hemophiliac males.

One affected X chromosome in a female makes her a "carrier" of hemophilia. A female hemophilia carrier may occasionally exhibit hemophilia symptoms. Find out more about the hemophilia inheritance pattern.

Despite the fact that hemophilia runs in families, some families have never had a relative with the disorder. But around one-third of the time, a mutation in the gene for the clotting factor affects the newborn that has hemophilia as the first member of the family to be afflicted.

#### Symptoms and signs

a) Bleeding into muscle and soft tissue, which results in bruising, or into the skin, which causes a blood clot in the area (called a hematoma).

b) Mouth and gum bleeding, as well as difficult-to-stop bleeding following tooth loss.

#### Diagnosis

Many people who have hemophilia or have relatives who do would request that their baby boys get tested as soon as possible after delivery.

When a baby is diagnosed with hemophilia, about one-third of them have a brand-new mutation that is absent in other family members. If a newborn exhibits certain hemophilia symptoms in these situations, a doctor may do a hemophilia test.

#### Treatment

In order for the blood to clot properly, replacing the missing blood clotting factor is the best strategy to treat hemophilia. By injecting commercially manufactured factor concentrations, this is accomplished. Hemophiliacs can learn how to administer these infusions by themselves in order to stop bleeding episodes and, by administering the infusions regularly, can even prevent the majority of bleeding events.

Some major issues can be avoided with high-quality medical care provided by professionals who are well-versed in the disorder, such doctors and nurses. The greatest option for care is frequently to go to a thorough hemophilia treatment center. An HTC offers health education to help persons with hemophilia maintain their health in addition to care for all conditions associated with the disorder.

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