Hemophilia

Dr.Narendra Kumar Garg
Professor-Kayachikitsa
Faculty of Ayurvedic Science
Jayoti Vidyapeeth Women's University, Jaipur

What is Hemophilia?

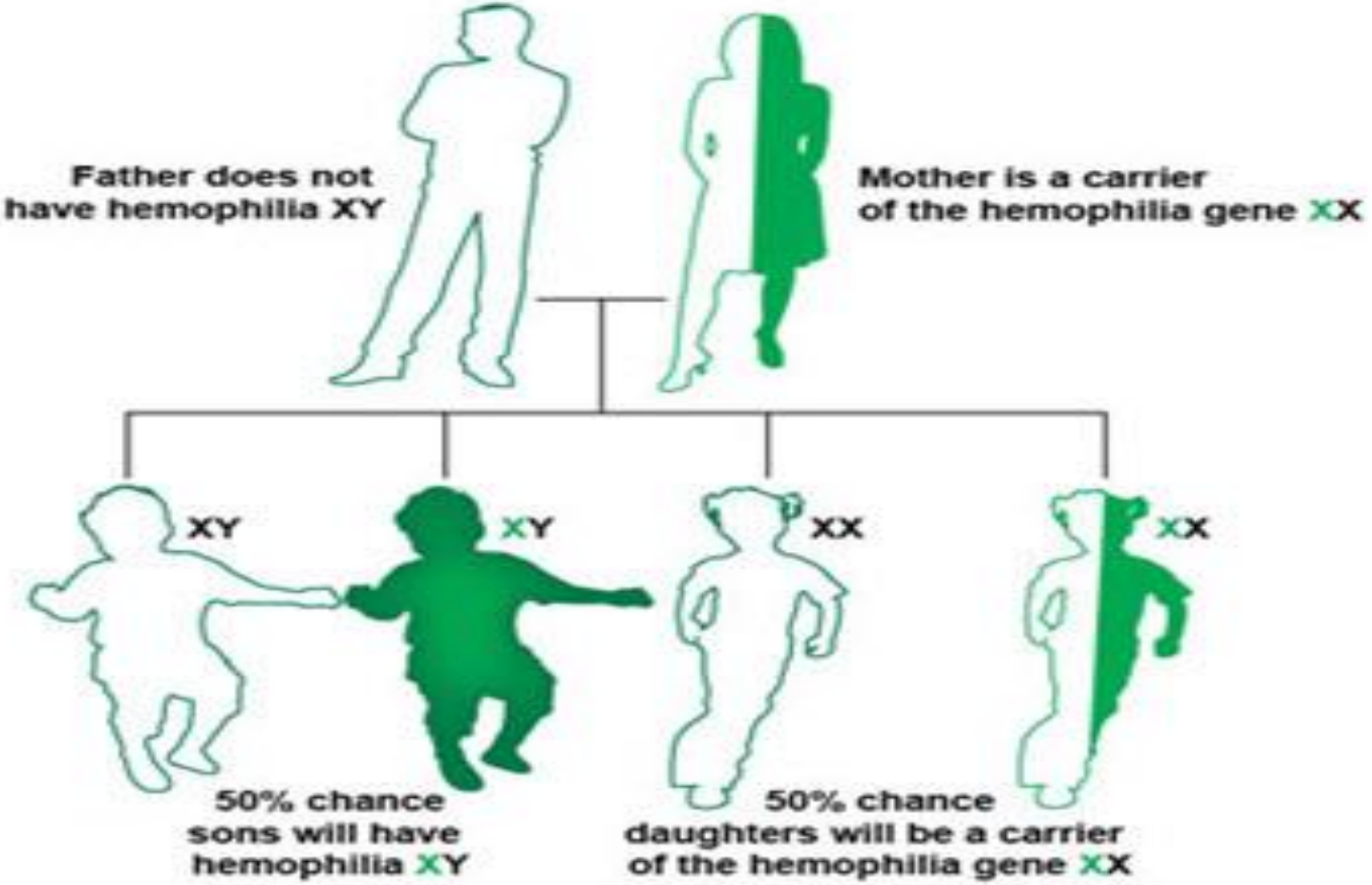
Hemophilia is usually an inherited bleeding disorder in which the blood does not clot properly. This can lead to spontaneous bleeding as well as bleeding following injuries or surgery. Blood contains many proteins called clotting factors that can help to stop bleeding. People with hemophilia have low levels of either factor VIII (8) or factor IX (9)

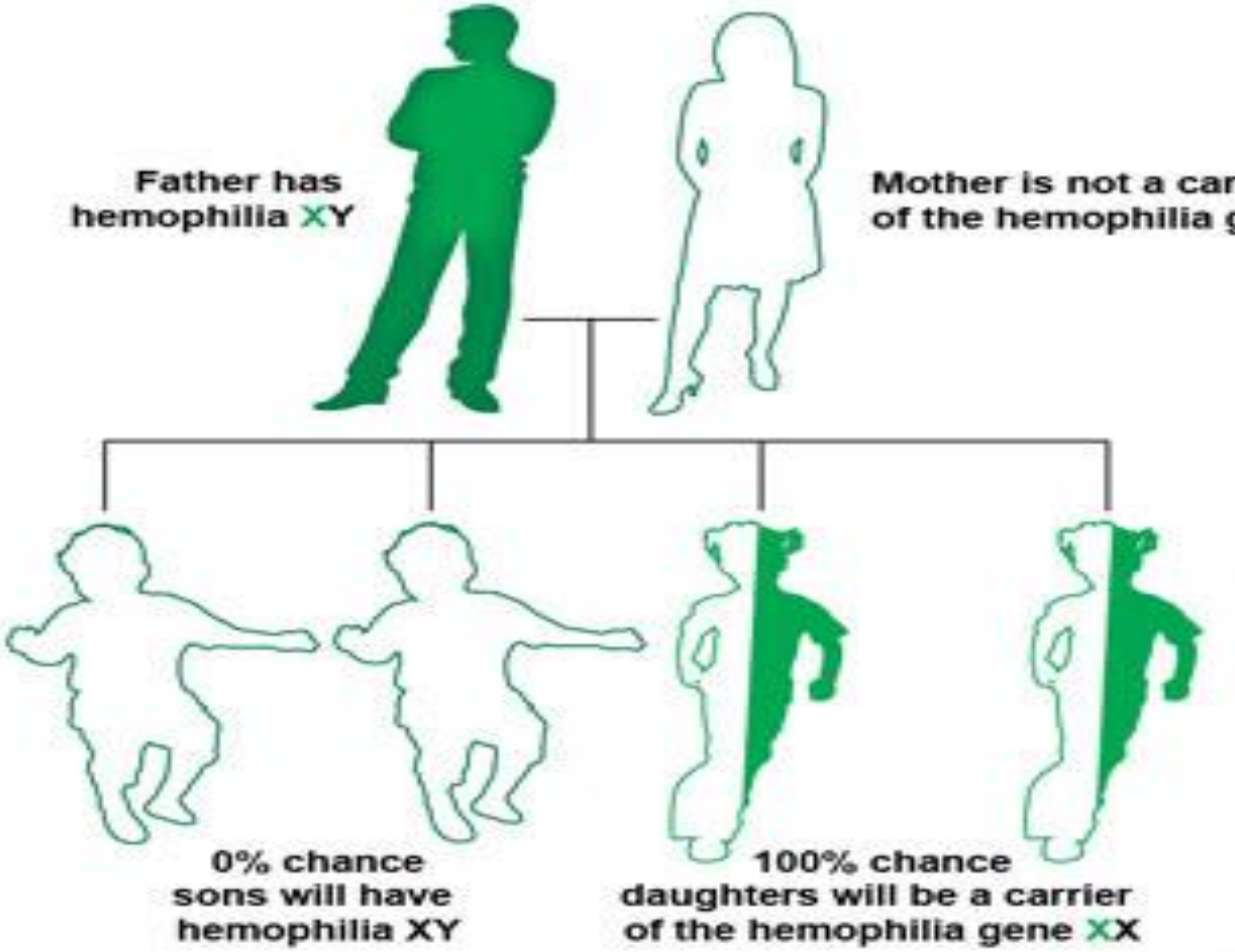
The severity of hemophilia that a person has is determined by the amount of factor in the blood. The lower the amount of the factor, the more likely it is that bleeding will occur which can lead to serious health problems.

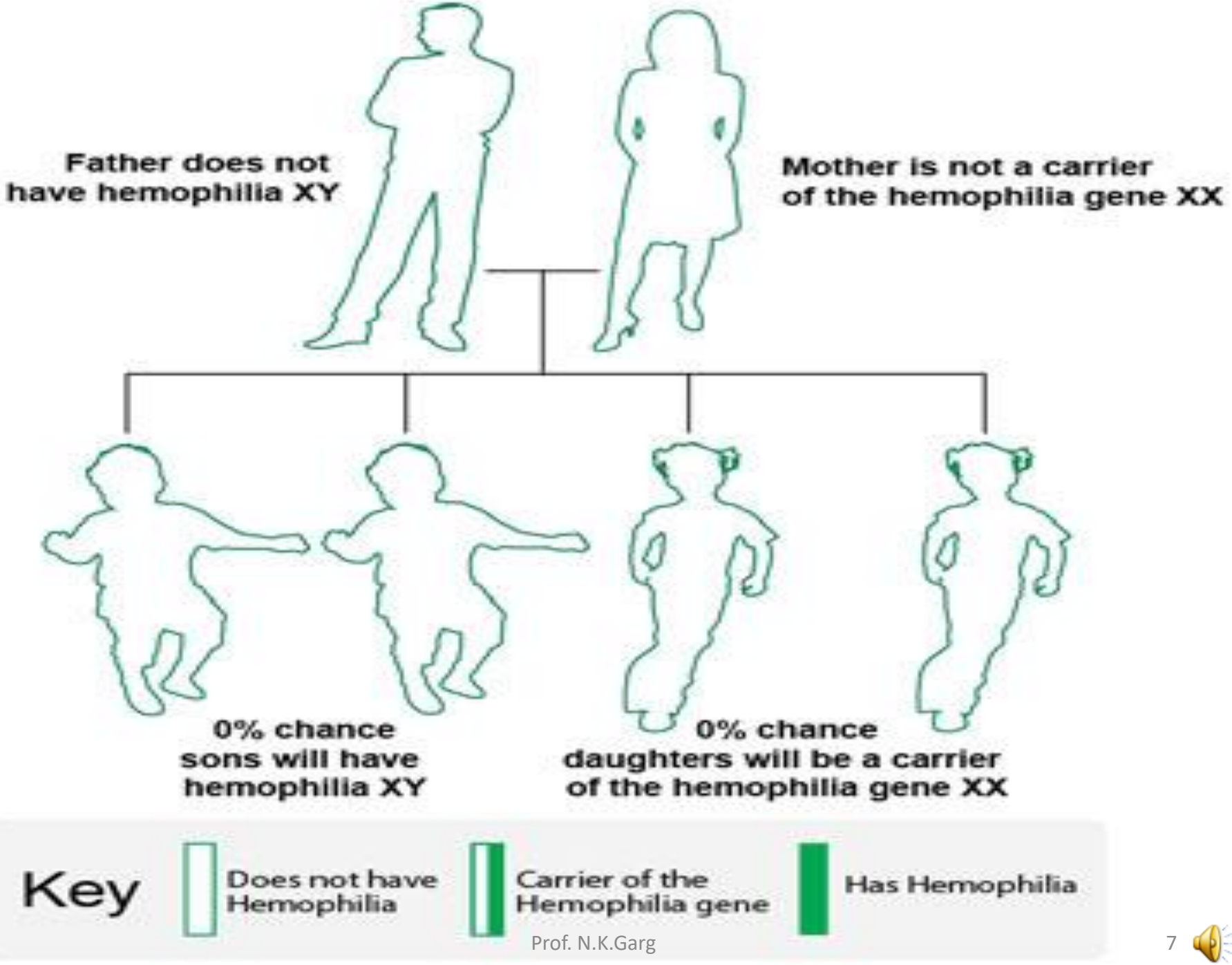
In rare cases, a person can develop hemophilia later in life. The majority of cases involve middle-aged or elderly people, or young women who have recently given birth or are in the later stages of pregnancy. This condition often resolves with appropriate treatment.

Causes

Hemophilia is caused by a mutation or change, in one of the genes, that provides instructions for making the clotting factor proteins needed to form a blood clot. This change or mutation can prevent the clotting protein from working properly or to be missing altogether. These genes are located on the X chromosome. Males have one X and one Y chromosome (XY) and females have two X chromosomes (XX). Males inherit the X chromosome from their mothers and the Y chromosome from their fathers. Females inherit one X chromosome from each parent.


Father does not have hemophilia XY
Mother is a carrier of the hemophilia gene XX
XY
XY
XX
XX
50% chance sons will have hemophilia XY
50% chance daughters will be a carrier of the hemophilia gene XX
Key
Does not have Hemophilia
Carrier of the Hemophilia gene
Has Hemophilia


Father has hemophilia XY
Mother is not a carrier of the hemophilia gene XX
0% chance sons will have hemophilia XY
100% chance daughters will be a carrier of the hemophilia gene XX
Key
Does not have Hemophilia
Carrier of the Hemophilia gene
Has Hemophilia


Father does not have hemophilia XY
Mother is not a carrier of the hemophilia gene XX
0% chance sons will have hemophilia XY
0% chance daughters will be a carrier of the hemophilia gene XX
Key
Does not have Hemophilia
Carrier of the Hemophilia gene
Has Hemophilia

The X chromosome contains many genes that are not present on the Y chromosome. This means that males only have one copy of most of the genes on the X chromosome, whereas females have 2 copies. Thus, males can have a disease like hemophilia if they inherit an affected X chromosome that has a mutation in either the factor VIII or factor IX gene. Females can also have hemophilia, but this is much rarer. In such cases both X chromosomes are affected or one is affected and the other is missing or inactive. In these females, bleeding symptoms may be similar to males with hemophilia.

A female with one affected X chromosome is a "carrier" of hemophilia. Sometimes a female who is a carrier can have symptoms of hemophilia. In addition, she can pass the affected X chromosome with the clotting factor gene mutation on to her children. Even though hemophilia runs in families, some families inheritance pattern for hemophilia have no prior history of family members with hemophilia. Sometimes, there are carrier females in the family, but no affected boys, just by chance. However, about one-third of the time, the baby with hemophilia is the first one in the family to be affected with a mutation in the gene for the clotting factor.

Types

There are several different types of hemophilia. The following two are the most common:

Hemophilia A (Classic Hemophilia)
This type is caused by a lack or decrease of clotting factor VIII.
Hemophilia B (Christmas Disease)
This type is caused by a lack or decrease of clotting factor IX.

hemophilia and the severity

Severity	Levels of Factor VIII (8) or IX (9) in the blood
Normal (person who does not have hemophilia)	50% to 100%
Mild hemophilia	Greater than 5% but less than 50%
Moderate hemophilia	1% to 5%
Severe hemophilia	Less than 1%

Diagnosis

Many people who have or have had family members with hemophilia will ask that their baby boys get tested soon after birth. About one-third of babies who are diagnosed with hemophilia have a new mutation not present in other family members. In these cases, a doctor might check for hemophilia if a newborn is showing certain signs of hemophilia.

To make a diagnosis, doctors would perform certain blood tests to show if the blood is clotting properly. If it does not, then they would do clotting factor tests, also called factor assays, to diagnose the cause of the bleeding disorder. These blood tests would show the type of hemophilia and the severity. Types of screening **tests** are

CBC - complete blood count .

APTT - activated partial thromboplastin time

PT - prothrombin time

Fibrinogen test.

Complete Blood Count (CBC)

The CBC is normal in people with hemophilia. However, if a person with hemophilia has unusually heavy bleeding or bleeds for a long time, the hemoglobin and the red blood cell count can be low.

Activated Partial Thromboplastin Time (APTT) Test

This test measures how long it takes for blood to clot. It measures the clotting ability of factors VIII (8), IX (9), XI (11), and XII (12). If any of these clotting factors are too low, it takes longer than normal for the blood to clot. The results of this test will show a longer clotting time among people with hemophilia A or B.

Prothrombin Time (PT) Test

This test also measures the time it takes for blood to clot. It measures primarily the clotting ability of factors I (1), II (2), V (5), VII (7), and X (10). If any of these factors are too low, it takes longer than normal for the blood to clot. The results of this test will be normal among most people with hemophilia A and B.

Fibrinogen Test

This test also helps doctors assess a patient's ability to form a blood clot. This test is ordered either along with other blood clotting tests or when a patient has an abnormal PT external icon or APTT test external icon result, or both. Fibrinogen is another name for clotting factor I (1).

Clotting Factor Tests

Clotting factor tests, also called factor assays, are required to diagnose a bleeding disorder. This blood test shows the type of hemophilia and the severity. It is important to know the type and severity in order to create the best treatment plan.

Signs and Symptoms

Common signs of hemophilia include:

- **Bleeding into the joints-** This can cause swelling and pain or tightness in the joints; it often affects the knees, elbows, and ankles.
- **Hematoma-** Bleeding into the skin (which is bruising) or muscle and soft tissue causing a build-up of blood in the area.
- **Bleeding of the mouth and gums-** bleeding that is hard to stop after losing a tooth.
- **Bleeding after circumcision** (surgery performed on male babies to remove the foreskin (covering the head of the penis).
- **Bleeding after having shots-** such as vaccinations.
- Bleeding in the head of an infant after a difficult delivery.
- Blood in the urine or stool.
- Frequent and hard-to-stop nosebleeds.

Who is Affected

Hemophilia occurs in about 1 of every 5,000 male births. Currently, about 20,000 males in the United States are living with the disorder. Hemophilia A is about four times as common as hemophilia B, and about half of those affected have the severe form. Hemophilia affects people from all racial and ethnic groups.

Treatment

The best way to treat hemophilia is to replace the missing blood clotting factor so that the blood can clot properly. This is done by infusing (administering through a vein) commercially prepared factor concentrates. People with hemophilia can learn how to perform these infusions themselves so that they can stop bleeding episodes and, by performing the infusions on a regular basis (called prophylaxis), can even prevent most bleeding episodes. Good quality medical care from doctors and nurses who know a lot about the disorder can help prevent some serious problems. Often the best choice for care is to visit a comprehensive Hemophilia Treatment Center (HTC). An HTC not only provides care to address all issues related to the disorder, but also provides health education that helps people with hemophilia stay healthy.

Inhibitors

About 15-20 percent of people with hemophilia develop an antibody (called an inhibitor) that stops the clotting factors from being able to clot the blood and stop bleeding. Treatment of bleeding episodes becomes extremely difficult, and the cost of care for a person with an inhibitor can skyrocket because more clotting factor or a different type of clotting factor is needed. People with inhibitors often experience more joint disease and other problems from bleeding that result in a reduced quality of life.

Do the 5"Key prevention messages from the National Hemophilia Foundation's National Prevention Program. Tips for Healthy Living

1. **Get an annual comprehensive checkup at a hemophilia treatment center.**
2. **Get vaccinated—Hepatitis A and B are preventable.**
3. **Treat bleeds early and adequately.**
4. **Exercise and maintain a healthy weight to protect your joints.**
5. **Get tested regularly for blood-borne**

To avoid excessive bleeding and protect your joints:

Exercise regularly. ...

Avoid certain pain medications. ...

Avoid blood-thinning medications. ...

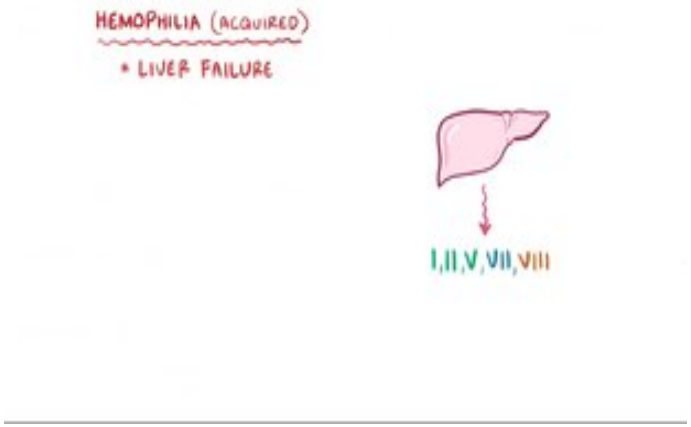
Practice good dental hygiene. ...

Protect your child from injuries that could cause bleeding.

Ayurvedic Management

Rakta is matrija bhava - this is raktapitta as kulaja vikara – Raktapitta treat. Should be apply eg. guduchi satva ,muktapishti and pravala pishti with vasa swaras. for longer duration if possible throughout the life


Hemophilia

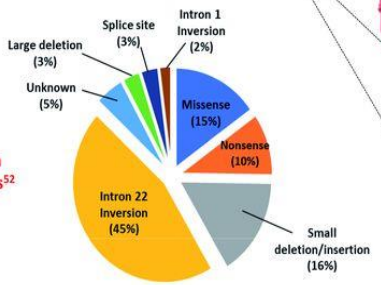

HEMOPHILIA (ACQUIRED)
• LIVER FAILURE
I,II,V,VII,VIII

Hemophilia A

Prevalence: 1:5,000 males

Mode of inheritance: X-linked recessive

Clinical symptoms: Joint bleeding, muscle hematoma, soft tissue bleeding



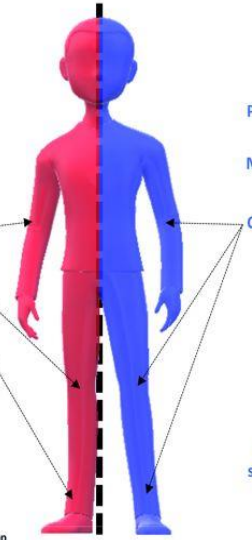
F8 gene defects reported in severe Hemophilia A patients[52]

Characteristics of missing clotting factor (FVIII):

Function: Co-factor

Molecular Weight: 280 kDa[53]

Normal concentration in plasma: 0.1-0.25 µg/mL

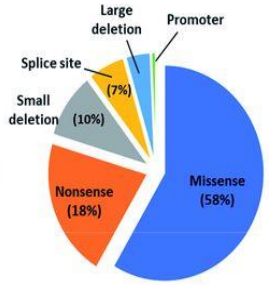


Hemophilia B

Prevalence 1:30,000 males

Mode of inheritance: X-linked recessive

Clinical symptoms: Joint bleeding, muscle hematoma, soft tissue bleeding



F9 gene defects reported in severe Hemophilia B patients[7]

Characteristics of missing clotting factor (FIX):

Function: Enzyme

Molecular Weight: 55 kDa[54]

Normal concentration in plasma: 3-5 µg/mL

Clot Formation

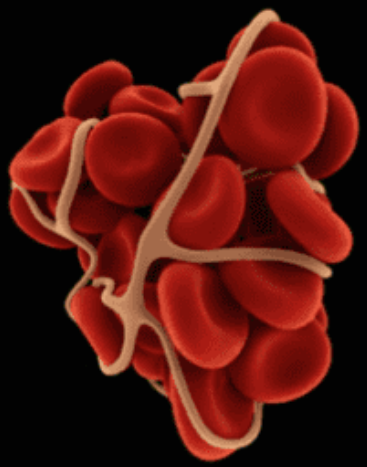
Primary Hemostasis

Platelets → Plug

Secondary Hemostasis

Clotting Factors → Fibrin

Support Clot → Glue




Hemophilia
askhematologist.com
Hemarthrosis-Right Knee

Haemophilia in the descendants of Queen Victoria

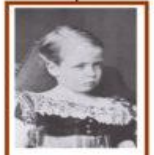
Queen Victoria
(1819-1901)

Princess Alice of the United Kingdom
(1843-1878)

Prince Leopold, Duke of Albany
(1853-1884)

Princess Beatrice of the United Kingdom
(1857-1944)

Princess Irene of Hesse and by Rhine
(1866-1953)



Prince Friedrich of Hesse and by Rhine
(1870-1873)

Alexandra Feodorovna of Russia
(1872-1918)

Princess Alice of Albany
(1883-1981)

Victoria Eugenie of Battenberg queen of Spain
(1887-1969)

Prince Leopold of Battenberg
(1889-1922)

Prince Maurice of Battenberg
(1891-1914)

Prince Waldemar of Prussia
(1889-1945)

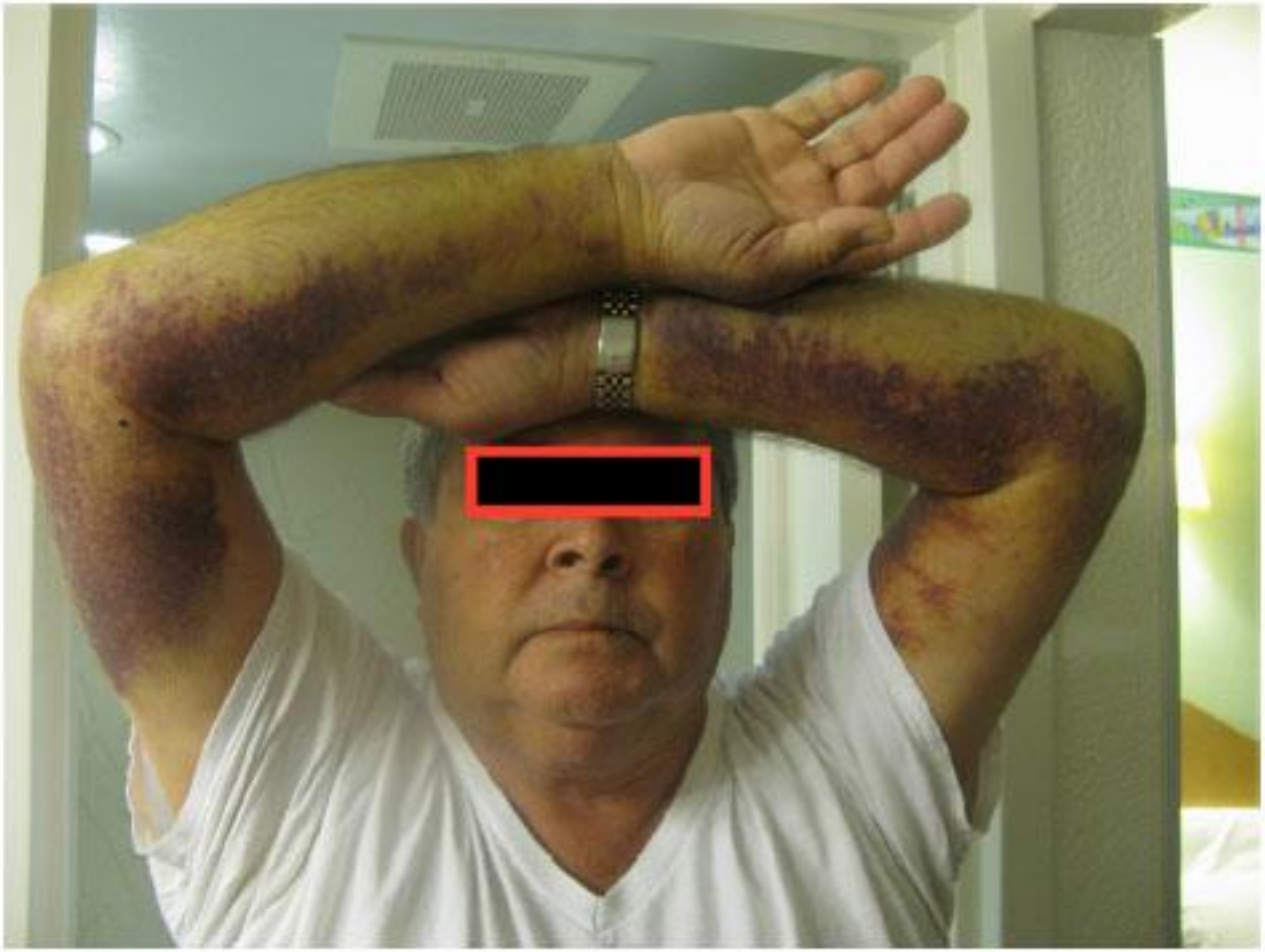
Prince Heinrich of Prussia
(1900-1904)

Alexey Nikolaevich of Russia
(1904-1918)

Prince Rupert of Teck
(1907-1928)

Alfonso of Spain, Prince of Asturias
(1907-1938)

Infante Gonzalo of Spain
(1914-1934)



Content source:
National Center on Birth Defects and Developmental Disabilities,
Centers for Disease Control and Prevention