HAEMOPHILIA

What is Haemophilia?

Haemophilia refers to a group of bleeding disorders in which it takes a long time for the blood to clot due to the absence or deficiency of clotting factors.

In most cases, the disorder is passed down through families (inherited).
- In Haemophilia A and B-it most often affects males but woman are the carriers of the defective genes.
- Von Willebrand disease affects men and women equally and mild bleeding may occur after surgery or when you have a tooth pulled.

Aspirin and other non-steroidal anti-inflammatory drugs (NSAIDs) can make this condition worse. Risk factors for Haemophilia in general are being male with a family history of bleeding. Genetic testing is available for concerned parents.

Rarely, adults can develop a bleeding disorder similar to Haemophilia A. This may happen after giving birth (postpartum), in people with certain autoimmune diseases such as rheumatoid arthritis, in people with certain types of cancer (most commonly lymphomas and leukemias), and also for unknown reasons (called "idiopathic"). Although these situations are rare, they can be associated with serious, even life-threatening bleeding.

What causes it?

- **Haemophilia A** - a lack of blood clotting factor VIII.
- **Haemophilia B** - a lack of blood clotting factor IX.
- **Von Willebrand disease** - a deficiency of von Willebrand factor. Von Willebrand factor helps blood platelets clump together and stick to the blood vessel wall. It is the least serious and most common hereditary bleeding disorder.

What are the symptoms?

The main symptom of hemophilia is bleeding. Mild cases may go unnoticed until later in life, when they occur during surgery or after trauma.

In more severe cases, serious bleeding may occur without any cause. Internal

bleeding may occur anywhere. Bleeding into joints is common.
Others symptoms may include:

- Blood in the urine or stool
- Bruising
- Gastrointestinal tract and urinary tract haemorrhage
- Nosebleeds
- Prolonged bleeding from cuts, tooth extraction, and surgery
- Excessive bleeding following circumcision
- Abnormal menstrual bleeding
- Bleeding of the gums

How is it diagnosed?

Most often, Haemophilia is diagnosed after a person has an abnormal bleeding episode or when there is a known family history of the condition.

If the patient is the first person in the family to have a suspected bleeding disorder, he or she will undergo a series of tests called a coagulation study. Once a disorder has been identified, other family members will need less testing to diagnose the disorder.

Blood tests to diagnose hemophilia A include:
- Low serum factor VIII activity
- Low factor IX
- Prothrombin time
- Bleeding time
- Fibrinogen level
- Partial thromboplastin time (PTT)
- Platelet aggregation test
- Platelet count
- Ristocetin co-factor test (the primary assay test used to diagnose von Willebrand disease)
- Von Willebrand factor level (level is reduced)

How is it diagnosed?

The outcome is usually good with treatment. Most people with haemophilia are able to lead relatively normal lives. However, some patients have significant bleeding events, most commonly chronic bleeding into the joint spaces which result in chronic joint deformities. This complication can be managed by an orthopaedic specialist. However, joint replacement may be needed.

Intracerebral haemorrhage is another possible complication. Repeated transfusions may slightly raise the risk for HIV and hepatitis , however, continued improvements in blood screening procedures makes blood products safer than ever.

Thrombosis may occur following use of factor IX concentrate. If you have Haemophilia and are scheduled for surgery or are in an accident, be sure you or your family notify the health care providers about your condition.

Treatment

Standard treatment is infusion of factor VIII or 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.

Mild Haemophilia A 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 Haemophilia A or B and their families can be taught to give factor VIII or IX concentrates at home at the first signs of bleeding. People with severe forms of the disease may need regular preventive treatment.

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.

Depending on the severity of the disease, factor VIII or IX concentrate may be given before having dental extractions or surgery in order to prevent bleeding.
Hepatitis B vaccine is recommended for individuals with Haemophilia B because they are at increased risk of developing hepatitis due to exposure to blood products.

Von Willebrand disease: Desmopressin (DDAVP) can be given to raise the levels of von Willebrand factor, which will reduce the tendency toward bleeding.

Blood plasma or certain factor VIII preparations may also be used to decrease bleeding.

Your role in managing this condition

Patients with Haemophilia should establish a good relationship with a haematologist, especially one who is associated with a haemophilia treatment centre. The ability to have quick and easy access to medical records documenting the patient's history of factor VII or IX levels, factor transfusions (including the type and amount), complications, and amount of any inhibitors can be lifesaving in the event of an emergency situation.

- Contact your health care provider if:
  - Symptoms of a bleeding disorder develop
  - A family member has been diagnosed with Haemophilia
- If you have Haemophilia, and you plan to have children, genetic counselling may be recommended.
- Testing can identify females who carry the Haemophilia gene. Prenatal intrauterine tests can be done to determine if a developing baby has the disorder.
- If you have von Willebrand disease, do not take non-steroidal anti-inflammatory drugs (NSAIDs), such as aspirin or ibuprofen, without talking to your health care provider.
- If you have Haemophilia and are scheduled for surgery or are in an accident, be sure you or your family notify the health care providers about your condition.
- Wear a Medic alert bracelet to notify caregivers about your condition.

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