وزارة الصحة
Ministry of Health

Hemophilia

Hemophilia:

Hemophilia is a genetic bleeding disorder that causes the blood not to clot normally because it lacks sufficient blood-clotting factors. People with hemophilia don't have as many clotting factors as there should be in the blood. This means that they bleed for a longer time than usual after an injury.

Definition of Coagulation (Blood Clotting):

Coagulation or blood clotting is a complex process that prevents excessive bleeding when a blood vessel is injured. This complex, multi-step process is done through a series of clotting factors (I-IX factors), and if any of these factors is missing or defective, it can lead to uncontrolled bleeding after an injury.

The Coagulation Process Includes the Following Steps:

- When an artery is injured or bruised, blood vessels quickly contract to help stop the bleeding.
- Small blood cells known as platelets immediately form a plug at the site of the injury to prevent blood from bleeding out of the injured blood vessel.
- At the same time, proteins in the blood plasma respond in a series of complex chemical reaction to form fibrin strands, which reinforce the platelet plug. In the meantime, the injured blood vessel begins to rebuild new cells instead of the damaged ones, and the temporary blood clot begins to vanish.

Types of Hemophilia:

Hemophilia is classified into three types based on the missing clotting factor:

- Hemophilia A: also called classic hemophilia, occurs when clotting factor VIII is either absent or not present in sufficient amounts.
- Hemophilia B: occurs when clotting factor IX is either absent or not present in sufficient amounts. It is the most common type in the Arab world.

- Hemophilia C: occurs when clotting factor XI is either absent or not present in sufficient amounts. It is very rare.

Causes of Hemophilia:

Hemophilia is caused by defects in the genes responsible for forming clotting factors. These genetic defects may be inherited from a parent, or caused by genetic mutations that develop during the formation of clotting factors during childhood, regardless of the fact that the parents do not have these defects.

Symptoms of Hemophilia:

- Unexplained and excessive bleeding either externally from cuts and injuries or internally, particularly in the muscles and joints. Bleeding may occur without any obvious triggering event or injury, or it may happen after minor surgeries (circumcision, tooth removal, etc.) or while being pricked by a needle for a blood test.
- One of the most serious types of internal bleeding is when the patient starts bleeding into the brain. This is usually accompanied by fainting or seizures. In such cases, the amount of bleeding depends

on certain factors including the patient's age, physical activity levels, and their clotting factor deficiency level.

- Newly walking or crawling infants start exhibiting blue bruises and injuries, especially on their knees, due to falling down frequently.
- Fibrosis and stiffness in the joints: repeated joint bleeds (hemarthrosis) often cause inflammation post-bleeding.
- Muscle impairment: after a few years of recurrent hemarthrosis and inflammations, the inflammatory process may lead to functional impairment causing the child to become physically disabled. If this child does not receive proper treatment at an early stage of the disease, then joint replacement surgery may become necessary upon reaching adulthood.
- In certain cases, when the disease is mild or moderate, symptoms may not appear until the patient undergoes surgery (e.g. tooth extraction, tonsillectomy, etc).

Hemophilia Is Diagnosed in Two Phases:

- **Blood tests are performed to determine how much factor VIII or factor IX is present**:
  - For Healthy individuals (with no hemophilia), the normal range of clotting factors in the blood is between ٥٠% and ١٠٠%.
  - Mild hemophilia: Individuals with mild hemophilia have between ٥% and ٥٠% of the normal number of clotting factors in their blood.
  - Moderate hemophilia: between ١% and ٥%
  - Severe hemophilia: Less than ١%
- **DNA test**:
  A blood sample is taken and then the DNA is extracted to test the gene responsible for forming clotting factors VIII and IX. The DNA

test can examine whether the gene is functioning properly, or if it is defective. This analysis takes three days at least.

Who Is at Risk of Hemophilia?

People of all nationalities, races, and ethnic groups can be affected by hemophilia. Most severe forms of hemophilia only affect males. In order for a female to develop severe hemophilia, the father must be a hemophiliac, and the mother a carrier of the disease; which is very unlikely to happen. Most women with the defective gene are simply carriers and experience no signs or symptoms of hemophilia, but some carriers can experience mild symptoms of the disease. Hemophilia is a genetic disease, and therefore, children are affected by it since birth.

Treatment:

Hemophilia treatment varies depending on the severity of the case:

- Mild hemophilia:
  The treatment of mild hemophilia involves slowly injecting desmopressin (DDAVP) into the vein to stimulate the production of more clotting factors to stop bleeding. Sometimes, the desmopressin drug is provided as a nasal spray.
- Moderate to severe hemophilia:
  Bleeding can be stopped only by getting a blood transfusion from a donor whose blood has the clotting factors the patient lacks, or by relying on genetically modified recombinant clotting factors. This process may have to be repeated in cases of internal bleeding.
- Severe hemophilia:
  Severe cases of hemophilia could be treated by transfusing plasma to stop frequent bleeding. Patients who have frequent bleeding episodes may be candidates for prophylactic factor infusions (regular infusion of clotting factor concentrates). These are given two or three

times per week to prevent bleeding from occurring.
The doctor must train the patient and members of the patient's family on how to administer a Desmopressin injection or a clotting factor injection at home, work, or school.

How to Treat Bleeding Caused by Injury or Tooth Extraction:

For oral bleeding, it is advisable to use a drug called Cyklokapron to prevent severe oral bleeding. This medication should only be taken for a limited period of time (٢-٨ days).
For injuries and internal bleeding (like a joint bleed), the first step is to stop the bleeding by applying first aid treatment measures, such as putting on a bandage, applying pressure to the area, and using ice packs. Then the clotting factor must be injected to stop the bleeding as fast as possible.

Comprehensive Hemophilia Treatment Centers:

Hemophilia is a potentially life-threatening disorder that can affect every aspect of a patient's life, as well as their families. In order to be able to address all angles of life with hemophilia, a new type of healthcare centers have emerged, providing comprehensive and specialized care for hemophilia patients. These centers offer:

- Several comprehensive healthcare services (physical and psychological care, dental care, orthopedic care, and general medical care).
- Extensive knowledge about the effects of psychological and social stress on mental and physical health.
- Patient support groups and other services that can help patients learn more about their disease and how to prevent it from affecting their daily lives. These centers also provide clotting factors and monitor their levels in the patient's body in case the patient needs to undergo surgery.

Coping with Hemophilia and Its Complications:

- The best way to avoid joint bleeds is by knowing, preemptively, when the bleeding is likely to occur, and taking the clotting factor in due time before the joint is affected.
- Use protective joint braces to protect your joints from injury.
- Cover ceramic, marble, or wooden floors with carpets and rugs. This reduces the risk of injuries, but it does not stop injuries from occurring altogether.
- Treating joint bleeds must be supervised by a medical team, so that they can provide the patient with the clotting factor as soon as possible.
- There are certain first aid procedures that hemophilia patients must follow in case of an injury. These procedures are named the "R.I.C.E." treatment, which is an acronym for basic first aid steps to treat injuries: rest, ice, compression and elevation.

Rest: Avoid moving the affected leg, or keep the affected arm in place to help with the recovery. If you continue to use the affected limb, you might risk further injury and bleeding in the muscles and joints.

Ice: Applying ice to the affected area can help increase the constriction of blood vessels. When blood vessels constrict, the flow of blood is restricted or decreased, which can help control the bleeding in the affected area. This process is called vasoconstriction.

Compression: Wearing a compression bandage on the injured joint can help support the joint and reduce bleeding.

Elevation: Raising the injured arm or leg above the level of the heart can reduce the amount of blood flowing to the injured area which slows down the bleeding.

After the initial treatment, when the bleeding stops and there is less pain,

swelling, and heat in the affected joint, the patient should undergo physiotherapy. It is very important to restore the joint's range of motion, which requires adhering to exercises prescribed by a physiotherapist. These exercises can help strengthen the muscles surrounding the affected joint, thus, protecting the joint from atrophy and stiffness, which in turn will reduce the frequency of spontaneous joint bleeds.

Clinical Health Education Department
For further questions kindly contact us via email:
Hpromotion@moh.gov.sa