Hemophilia

Bleeding is a natural response to wounds on any part of the body, but a healthy blood system should eventually clot to stop the bleeding. This intricate process of blood clotting relies on platelets and 13 essential proteins known as clotting factors. These factors play a crucial role in coagulation, and even a slight reduction in their levels can result in slow or insufficient clot formation, leading to excessive bleeding.

Hemophilia, a bleeding disorder, is primarily characterized by deficiencies in Factor 8 (known as hemophilia A) and Factor 9 (known as hemophilia B). The genes responsible for producing these factors are located on the X chromosome. In females, who possess two X chromosomes, the presence of a defective gene on one chromosome is often compensated by a healthy gene on the other, making this disorder less common in women. However, for males with only one X chromosome, a defect in the corresponding gene leads to a deficiency in the factor, causing hemophilia. These gene abnormalities, known as mutations, can be inherited and are passed down through generations.

With approximately 1000 identified mutations in factor genes, hemophilia affects roughly 1 in every 5,000 to 10,000 male children. Despite its prevalence, awareness of this condition remains disappointingly low, and many individuals suffer and even succumb to the disease without proper diagnosis.

The severity of hemophilia depends on the extent of the factor deficiency. A classification system categorizes cases with factor levels above 5% as mild, between 1% and 5% as moderate, and less than 1% as severe. Patients with mild and moderate forms tend to experience fewer complications compared to those with severe hemophilia. While individuals with severe hemophilia may encounter severe bleeding even with minor injuries, those with milder forms usually have bleeding issues only in response to more significant traumas such as tooth extraction, surgeries or accidents.

Hemarthrosis, a type of bleeding, commonly affects the knee joints. It can manifest for the first time when a child starts walking, typically around one and a half years old. When bleeding occurs in the joint, it leads to swelling, redness, and intense pain. Moreover, repeated bleeding in the same joint can cause further complications. Over time, the joint's shape may change, mobility decreases, and the surrounding muscles weaken, raising the risk of subsequent bleeding episodes. The joint which gets involved with such complication is known as the "target joint". In some cases, bleeding may occur into the muscles, forming lumps. Additionally, head injuries can trigger bleeding in the brain, increasing the potential for severe consequences, including a high risk of mortality.

To confirm the presence of hemophilia, blood tests are conducted in two stages. Initially, platelet count, PT, and APTT tests are performed. In hemophilia patients, the APTT test results are notably elevated. Subsequently, the amount of factors is determined based on the APTT results. These tests must be carried out immediately after collecting the blood sample. Hence, it is recommended to conduct these tests in specialized laboratories. Although blood samples can be sent by courier, there is a chance of inaccurate test results in such cases.

Similar to providing hormone pills for individuals with thyroid hormone deficiencies, patients with severe factor deficiencies require factor replacement therapy to manage hemophilia effectively. In the management of hemophilia, factors play a crucial role as they are given both during bleeding episodes and as a preventive measure, known as prophylaxis. Prophylaxis aims to ensure that the patient never experiences bleeding and achieves proper physical and mental development.

In the past, factors were expensive, making this form of treatment financially burdensome. However, the scenario has changed, and factor prices are gradually becoming more affordable. Moreover, newer factor replacement therapies, such as Emicizumab, are now available, further expanding treatment options. These advancements indicate that prophylaxis to prevent bleeding may become widely accessible to all patients in the near future.

Prompt administration of factors is vital as soon as bleeding occurs or is suspected. Immediate intervention helps to stop bleeding swiftly and effectively, reducing the risk of joint damage and complications for the patient. The dosage and frequency of factor administration depend on the location and severity of the bleeding, and this decision is made by the treating doctor. In situations where factors are unavailable, cryoprecipitate or plasma transfusion can be alternative treatment options to manage bleeding episodes effectively.

The RICE regimen is a highly effective and widely recommended approach for managing bleeding episodes in individuals with hemophilia. Following this regimen for 24 hours after bleeding can significantly aid in reducing pain, swelling, and the duration of the bleeding episode. Let's break down the components of the RICE regimen:

R - REST: Complete rest is crucial for the bleeding part of the body. For joint bleeds, using a walker for walking during the initial week is advisable. After a week, gradual exercise therapy can be started to promote joint mobility and strengthen the muscles.

I - ICE: Applying ice to the bleeding joint can help constrict blood vessels, reducing blood flow to the affected area. To do this, wrap ice in a plastic bag, cover it with a cloth, and apply it to the bleeding joint for 20 minutes. After this, leave the joint undisturbed for three hours. The intermittent application of ice helps minimize inflammation and pain.

C - COMPRESSION: Applying gentle compression to the bleeding joint with a crepe bandage can effectively halt bleeding. The pressure applied through compression aids in controlling bleeding and reduces the extent of swelling.

E - ELEVATION: Elevating the bleeding joint above the level of the heart helps in two ways. Firstly, it reduces blood flow into the joint, limiting the accumulation of blood. Secondly, elevating the joint facilitates the draining of fluid away from the affected area, leading to a reduction in swelling and pain.

In addition to the RICE regimen, specific pain relievers are recommended for individuals with hemophilia. Celecoxib and Paracetamol are two such pain relievers that can be safely administered. However, it's essential to avoid other analgesics, as they can increase the risk of bleeding. Furthermore, the use of tranexamic acid tablets can play a crucial role in stopping bleeding episodes and may be prescribed as part of the treatment plan.

People with hemophilia should never recieve intramuscular (IM) injections due to the risk of excessive bleeding. Engaging in activities where injury is possible should also be avoided. However, they can safely participate in low-impact sports like racket games and swimming. Keeping factor injections readily available at home is essential for these patients. It is crucial for both the patient and patient's parents to be knowledgeable about calculating the injection dose, as well as preparing and administering the injection.

Physiotherapy treatment plays a vital role in maintaining the health of individuals with hemophilia. Strengthening the muscles can reduce the likelihood of bleeding incidents. Therefore, patients with hemophilia should never neglect prescribed physiotherapy exercises provided by their physiotherapist. Swimming is an excellent exercise for such patients and should be incorporated into their regular routine.

Administering hepatitis vaccines to hemophilia patients is crucial in preventing hepatitis development.

In cases where the knee joint has severely deformed due to frequent bleeding, joint replacement surgery may be considered. However, such surgeries require a high amount of factors during the procedure and post-surgical physiotherapy treatment.

In some cases, patients may develop antibodies, known as inhibitors, against the external factors administered to control bleeding. These inhibitors render the injected factors ineffective, making their administration during bleeding futile. In such situations, alternatives like FIEBA or Recombinant Factor 7 (Novoseven) injections can be given to stop bleeding, and Emicizumab injections can be utilized to prevent bleeding in patients with inhibitors.

As previously mentioned, hemophilia is a genetic disorder caused by a defect in the factor's genes. Treatment options are now available in western countries to replace these defective genes, enabling patients to produce sufficient factors by themselves, allowing them to lead normal lives.

Clinical trials, including gene therapy, are also underway in India. Patients who have the opportunity to participate in these trials should consider taking advantage of such opportunities.

As hemophilia is a hereditary disease passed down through generations, genetic testing and counseling of relatives should be carried out. There is a possibility

that the patient's sister is a carrier, and her sons might become hemophilia patients. Preventing this by genetic testing is possible now in India and the patient's family should make use of these facilities.

It is important to note that hemophilia is not contagious. There is no requirement for these patients to follow any special diet.

For any further questions or concerns about hemophilia, beyond the information provided here, patients are advised to consult their treating physician

Dr. Girish Kamat MD, DNB (Hematology)
Professor,
Department of Hematology,
SDM College of Medical Sciences and Hospital,
Sri Dharmasthala Manjunatheshwara University,
Dharwad- 580008