Hemophilia essay

Hemophilia is a genetic bleeding disorder. People who have hemophilia have a deficiency or an absence of a coagulation protein. A blood clotting factor is deficient or absent. Bleeding is most often into joints, such as the knee, elbow, or ankle, but bleeding can occur anywhere in the body. People with hemophilia bleed longer, not faster.

The severity of hemophilia varies greatly. Hemophilia A and Hemophilia B are the most common genetic bleeding disorders. Hemophilia A is observed in 80 percent of hemophiliacs and is a deficiency or absence of Factor VIII. It can also be referred to as “ classic” hemophilia.

In the second most common, hemophilia B, factor IX is missing. This is also known as the “ Christmas Disease” because of the surname of the first patient studied. Hemophilia was identified as early as biblical times. Doctors in medieval times were familiar with it as well. In 1803, a Philadelphia doctor published the first description of hemophilia in the United States.

But it was not until 30 years later that hemophilia became widely recognized.

Hemophilia later developed a reputation as the “ royal disease” because it passed from Queen Victoria of England to her descendants throughout the royal houses of Europe. About eighty percent of all cases of hemophilia have an identifiable family history of the disease; in other instances, it may be attributable to a spontaneous mutation. Researchers recently discovered that the spontaneous mutation of the factor VIII gene in two children was due to the attachment of a foreign “ jumping gene” that disrupted the blood-clotting ability of the factor VIII gene.

Inheritance is controlled by a recessive sex-linked factor carried by the mother on the X chromosome. A probability of one in two exists that each boy born to a normal male and a carrier female will be hemophiliac and the same chance that each girl of this union will be a carrier. Of the children of a hemophiliac male and a normal female, all the girls will be carriers and all the boys will be normal.

Males cannot transmit the disability, and female carriers are free of the disease. Conventional wisdom suggests that 1 in 10, 000 males in the United States have hemophilia.

However, increased research and focus, on bleeding disorders in general and on bleeding disorders in women specifically, suggest a shift in what is known about who has a bleeding disorder. Clotting factor is one of 12 or more proteins found in blood that work together to make blood clot. They are designated by Roman numerals I through XIII. When the body detects bleeding, clotting factors are switched on in a specific order, each sending an activating message to the next. Factor VIII is one of the clotting factor proteins that helps produce the fibrin clot.

Sufficient quantities of fibrin must be made by the body in order for fibrin to act like a net that holds the platelets together to make a firm blood clot. In persons with hemophilia A, fibrin is not made properly, so firm blood clots do not form in the wound, and bleeding continues. There are two major processes involved in blood clotting. The first part has to do with platelets.

They are like little shingles which go to where a blood vessel has ruptured, and they stick over the hole and make a plug.

This is the first step of making a clot. The plug is only temporary, and the platelets can easily fall off. The platelets soon rupture and release chemicals that attract more platelets and make them “ sticky”, too. The chemicals released by the rupturing platelets also activate various clotting factors which are proteins in the blood.

The next step is that fibers form from the activated proteins and mix with the platelets. The fibers are like a net, or a weave of yarn, and they make the clot stronger. The substance that makes the fibers is called fibrinogen. There are twelve factors which work together to make the fibrinogen. People with hemophilia have a problem with one or more of those factors.

The most common of the twelve factors to have a problem is factor VIII, which causes hemophilia A.

The second most common to have a problem is factor IX, and this causes hemophilia B. Babies with hemophilia usually have no difficulty during the birth process, however circumcision may produce prolonged bleeding. During the first few months of life, a baby with hemophilia has few problems because ones ability to move around is limited. As the baby learns to walk one will fall and sustain many small, superficial bumps and bruises. Bleeding into soft tissue area of the arms and legs is frequent and usually not serious.

Superficial bruises are seen easily and are often raised, but usually do not require treatment. As a child with hemophilia matures, one is more active and will have more bleeding episodes. The first sign of bleeding deep in a muscle may be a reluctance to use the limb. The child may become irritable as bleeding continues.

When the symptoms are present, examination of the limb should be done carefully but do not in a manner where the limb should be moved forcibly is one resists. After an examination the opposite limb at the same time has been completed, one can detect possible differences in swelling and/or skin temperature.

It is crucial to also beware of a complaint from an older child of a pulled muscle". Those are often confused with deep muscle bleeding. A pulled muscle complaint is always to be considered a deep muscle bleeding unless proven otherwise. A bleeding episode in a muscle can spread through the muscle length, often without one noticing it.

When muscle bleeding occurs in the forearm, calf or groin, swelling can create pressure on the nerves which can result in numbness, and subsequent pain and inability to move the limb. Deep muscle bleeding often requires follow-up treatment and observation by the center staff. Sling, splints or crutches may be necessary for support for several days. As one with hemophilia grows, bleeding into his joints will occur more frequently.

Early signs of bleeding are the reluctance to use the affected limb followed by a slight swelling in the affected joint.

Attempts to move the joint forcibly can cause pain. As more blood leaks into the joint it feels warmer than the opposite one and the swelling feels " spongy". Later the person holds the limb in a bent or " flexed" position to ease pain. Flexion increases the space in the joint, making more room for further bleeding to occur. If untreated, the bleeding continues until the area feels hot and rock hard.

Usually there is no bruising associated with a joint bleed, and since the bleeding occurs in an enclosed space (the joint capsule) and has nowhere to spread, the pressure caused by the bleeding eventually results in pain — which can be quite severe. Early treatment with clotting factor usually prevents the pain from becoming severe, and since only a small amount of blood will have leaked into the joint space, recovery will be quick, although the joint will sustain some permanent damage. In addition to pain considerations, inadequately treated bleeding irritates the joint surface which eventually leads to arthritis.

A large volume of blood, which results from delayed treatment, takes longer to re-absorb into the body.

The longer the period of irritation, the greater the chance of arthritic damage. Early treatment of each bleeding episode will limit the amount of irritation and reduce the risk of arthritis. Strong muscles protect joints from some bleeding episodes. Therefore it is suggested that people with hemophilia exercise regularly. Bleeding in the mouth can be troublesome and messy, but is usually minor as long as there is no swelling of, or bruise, under the tongue.

Blood mixed with saliva may make the bleeding look much worse than it actually is. Don’t panic. Older persons with hemophilia will know when they are bleeding, long before there is any externally noticeable symptom. While it does depend on the individual, one can expect a grown person with hemophilia to manage their own condition and treatment effectively in most cases.

Prevention of injury is important for the patient with hemophilia. When bleeding occurs, replacement therapy may be necessary. Current products used to treat hemophilia are either manufactured from fresh frozen plasma, which are from single blood donors and require special freezing, or are “ freeze dried” factor VIII and factor IX concentrates. These concentrates are made in large lots, come in small bottles and may be kept at room temperature or in the regular refrigerator.

Some of the concentrates are made from large pools of donated plasma, and the newest and most expensive are made using recombinant gene technology. The recombinant factor is not made from human blood, however, it is manufactured using some human blood components, and other animal biological components.

There is still risk involved in treatment with any of these products. However, not treating can be very painful, and lead to other serious problems.

All blood donors in the U. S. are tested for blood borne viruses and all blood products are tested for hepatitis and the virus that causes AIDS. In addition, concentrates are treated and purified in several ways to reduce the risk of hepatitis and virtually eliminate the risk of AIDS transmission. Monoclonal products are more pure than heated concentrates, and recombinant factor is considered the most pure. All factor replacement products are done intravenously.

When clotting factor is administered the body begins to use it right away to form a clot. After 6-8 hours the body has “ used up” about half of what was administered. Usually by this time most bleeding is well controlled.

After about 24 hours the body has essentially used up all the clotting factor, at which time the clot is well established and the body begins to get rid of the blood which has leaked into the surrounding tissues, so that eventually the swelling subsides.

There are two important points to keep in mind: A fresh bleeding episode can start if the clot becomes dislodged and natural reactions in the body cause a clot that is no longer needed to “ break down” or to be dissolved. The processes begin approximately five days after the initial clot was formed and occasionally a bleed may restart at the site. Although proficient, sometimes one infusion is not enough and one must be prescribe a follow up treatment. The most significant advances in hemophilia treatment have been made in the last four decades.

Baxter Healthcare Corporation introduced the first commercially available plasma-derived factor concentrate in the mid-1960s.

This was a major advancement over earlier formulations, which contained much lower concentrations of antihemophilic factor. In the early 1970s, home treatment of hemophilia became widely available, offering people with hemophilia greater independence and reduced hospital stays. Today, recombinant DNA technology and the discovery of the genes that control production of factor VIII have led to the development of recombinant factor concentrates that do not rely on plasma at all. Patients should discuss with their physicians which factor VIII replacement therapy is best for them. Inhibitors are proteins called antibodies that are made by our immune system to defend us from harmful disease.

When our immune system identifies a foreign substance, it makes antibodies that will specifically recognize that substance and destroy it. In some individuals with severe hemophilia, the factor VIII replacement therapy is identified as a foreign substance by their immune system. If this happens, their immune system will make antibodies against factor VIII. These antibodies will inhibit the ability of the factor to work in the clotting process.

The higher the antibody or inhibitor level, the more factor VIII replacement therapy it takes to overcome the inhibition and produce clotting. This can complicate the treatment of a bleed. The good news is that there are different types of therapies available to successfully treat most individuals who develop inhibitors.

Bibliography:

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