

Genetic disorder



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Introduction: At one time or another in life, one has cut himself. When that happens, there is bleeding immediately and then blood clots stopping the blood flow from the injury. Hemophilia is a condition where blood does not clot normally and it takes a long time to stop the blood flow. Hemophilia is life threatening when a person has a serious injury (Cutler, para1-2).

Hemophilia is a genetic disease and it results from a mistake or defect in one of a person's genes. Genes are components in our body cells that carry the information that establish our traits. Therefore, in the case of hemophilia, the gene that controls the making of substances that cause blood clotting has a defect. It is possible for parents to pass this problem to their children.

According to the World Federation of Hemophilia statistics, there are more than 500, 000 people with hemophilia worldwide (Freedman, p45-46).

Blood clotting is a result of chemical activities in the body. When one is wounded, the blood vessels are affected. Platelets clump together to begin plugging the hole. This process is called platelets adhesion. The first platelets to reach the injured area block the chemicals that draw proteins called clotting factors. As soon as the clotting factors get into the scene, they assist in the formation of fibrin, a chain of proteins. The threads of fibrin form a web of tough fibers around the platelets, holding them tightly in place.

When one has hemophilia, one of the clotting factors is lacking or is not working properly. The blood clot forms are normally soft and can easily fall apart (Freedman, p48).

There are two types of Hemophilia namely; hemophilia A and hemophilia B, which is the most common and both types result from a faulty gene that impedes the production of the clotting factors that permit the blood's normal ability to clot resulting in unusual, acute bleeding tendency. Both forms of

hemophilia may either be mild whereby bleeding only happens under severe stress for instance major injury, moderate where spontaneous bleeding is rare but bleeding occurs after trauma or surgery or severe whereby spontaneous bleeding is common. In spontaneous bleeding, a person bleeds in any body part especially in the spine finger-joints, feet and wrists, even with no recognizable trauma (Cutler, para2).

Hemophilia is more common in males than females. The scientific explanation is that the gene for hemophilia is found in X chromosome. X chromosome determines the sex of a baby. Females have two X chromosomes while males have one Y chromosome and one X chromosome. Babies get the X chromosomes from their mother and Y or X chromosome from their father. The gene for hemophilia is called X-linked recessive gene. If a male X chromosome has a defective gene for one of clotting factors, he will have hemophilia. However, if a female receives an X chromosome from one parent that has defective gene for the clotting factors and she gets a healthy gene for the same factor on the X chromosome from the other parent, she will not get hemophilia.

A female who is carrying a defective gene in her X chromosome is a carrier. Carriers often have other related setbacks, even though they do not have hemophilia. For example, they may have abnormal bleeding during surgery, menstruation or nose bleeding. When pregnant, the child of a carrier has a 50 percent chance of inheriting the defective gene. If it is a son, he will receive the gene with hemophilia but if it is a daughter, she will be a carrier (Freedman, p54).

Conclusion

The total treatment of hemophilia patients consists of providing adequate

supply of missing blood clotting factor at the time of bleeding and medical attention of qualified team of physicians. Each patient requires an individualized treatment program with correct orthopedic and medical examinations to determine the best course of therapy (Dietrich, p343).

Home treatment on the other hand forms an established part of medical care for hemophilia patients and is administered by hemophilia treatment centers nationwide and individual physicians. Criteria for choosing of the patient who can have home treatment include absence of medical complications, severe factor VIII or IX deficiency and accessible veins.

Another most important factor is the willingness and motivation on the part of the patient to learn more about the condition. The number of lessons required depends with the ability of the patient, the parent or wife to learn the right techniques. Home treatment is important, especially to those in school and at work, as the ability to treat it minimizes absenteeism from school and work (Dietrich, p345).

Works Cited

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