Hemophilia

**Hemophila, **<<hee muh FIHL ee uh,>> is a hereditary disease in which the blood does not clot normally. People with hemophilia, called hemophiliacs, bleed excessively when injured because their blood clots extremely slowly. Almost all hemophiliacs are males.

Blood must contain a number of substances called **clotting factors** for clotting to take place. A hemophiliac's blood lacks the active form of one of these factors. Hemophiliacs suffer most when internal blood vessels break, causing bleeding into such areas as the head or joints. Leaking blood accumulates in these areas, putting pressure on the surrounding tissues and causing pain, swelling, and loss of function. Many hemophiliacs become crippled from repeated bleeding into joints. Some people believe that hemophiliacs can bleed to death from even a small external cut. However, such deaths do not occur because blood contains clotting factors that halt bleeding from the skin.

**How hemophilia is inherited.** Hemophilia is caused by a defective gene on the **X chromosome**, one of the two chromosomes that determine a person's sex (see Heredity (Patterns of heredity)). The **Y chromosome**, which has no genes for clotting factors, is the other. Males have one X chromosome and one Y chromosome. Females have two X chromosomes.

A boy who inherits the hemophilia defect on his X chromosome will be a hemophiliac. A girl who inherits the defective gene on one of her X chromosomes will be a **carrier**. She may transmit the defective gene to her children. But she will not be a hemophiliac because the normal gene on her other X chromosome provides enough of the essential clotting factor. In extremely rare cases, a girl inherits the defective gene on both X chromosomes and will be a hemophiliac.

**Major types of hemophilia** include classical hemophilia and Christmas disease. The name of Christmas disease comes from the last name of one of the first patients to be treated for it. About 85 per cent of all hemophiliacs have classical hemophilia. Their blood lacks a protein called **clotting factor number 8**. Nearly all others have Christmas disease, which involves the lack of **clotting factor number 9**. An extremely small number of hemophiliacs lack another kind of clotting factor.

A blood disorder called **von Willebrand's disease** is sometimes confused with hemophilia. Unlike hemophilia, von Willebrand's disease causes prolonged external bleeding and affects both sexes. But few people with this blood disorder suffer bleeding into the joints.

**Treatment** for hemophilia consists of injections of the clotting factor that is lacking in the blood. The injections, which are made from donated blood, temporarily cause normal clotting. Treatment should be given soon after an injury, so that blood does not accumulate and thus damage body tissues. Many hemophiliacs keep a supply of clotting factor and inject themselves.

During the early and mid-1980's, hundreds of hemophiliacs became infected with HIV, the
virus that causes AIDS (acquired immunodeficiency syndrome), after receiving clotting factor contaminated with the virus. Since 1985, blood banks process clotting factor from donated blood to destroy the HIV virus, greatly increasing the safety of treatment. In addition, clotting factor produced in the laboratory through techniques of genetic engineering is available for hemophilia treatment.

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