

Hemophilia A

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Hemophilia A is a hereditary blood disorder, primarily affecting males, characterized by a deficiency of the blood clotting protein known as Factor VIII that results in abnormal bleeding. Babylonian Jews first described hemophilia more than 1700 years ago; the disease first drew widespread public attention when Queen Victoria transmitted it to several European royal families. Mutation of the HEMA gene on the X chromosome causes Hemophilia A. Normally, females have two X chromosomes, whereas males have one X and one Y chromosome. Since males have only a single copy of any gene located on the X chromosome, they cannot offset damage to that gene with an additional copy as can females. Consequently, X-linked disorders such as Hemophilia A are far more common in males. The HEMA gene codes for Factor VIII, which is synthesized mainly in the liver, and is one of many factors involved in blood coagulation; its loss alone is enough to cause Hemophilia A even if all the other coagulation factors are still present. Treatment of Hemophilia A has progressed rapidly since the middle of the last century when patients were infused with plasma or processed plasma products to replace Factor VIII. HIV contamination of human blood supplies and the consequent HIV infection of most hemophiliacs in the mid- 1980s forced the development of alternate Factor VIII sources for replacement therapy, including monoclonal antibody purified Factor VIII and recombinant Factor VIII, both of which are used in replacement therapies today. Development of a gene replacement therapy for Hemophilia A has reached the clinical trial stage, and results so far have been encouraging. Investigators are still evaluating the long-term safety of these therapies, and it is hoped that a genetic cure for hemophilia will be generally available in the future. For Diagnosis and treatment [click here](#) Important Links
Gene sequence

LocusLink [www.ncbi.nlm.nih.gov/LocusLink/list.cgi?Q=hemophilia ORG=Hs V=0] collection of gene-related information

BLink [www.ncbi.nlm.nih.gov/sutils/blink.cgi?pid=4503647 org=1]

OMIM [www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=306700]