

Hemophilia a genetic disorder: diagnosis, treatment and prognosis

Abstract

Hemophilia is a genetic disorder with patient's inability to stop bleeding. There are two main types of hemophilia, hemophilia A due to not enough clotting factor VIII and hemophilia B due to not enough factor IX, and acquired hemophilia A (AHA) caused by autoantibodies against clotting factor VIII (FVIII). AHA is associated with malignancy, autoimmune disorders, and pregnancy. Factor IX deficiency can cause interference of the coagulation cascade. People with more severe hemophilia usually suffer more severe and more bleeds than people with mild hemophilia. Complications of hemophilia include deep internal bleeding, joint damage, transfusion induced infection, adverse reactions to clotting factor treatment, and intracranial hemorrhage. Diagnosis of hemophilia can be confirmed by, coagulation screening test, bleeding scores and coagulation factor assay. Gold standard of treatment is rapid treatment of bleeding episodes decreases damage to the body. Prophylactic treatment although high costs, is more effective than on demand treatment. People with severe hemophilia without adequate treatment have generally shortened lifespans. Gene therapy is not currently an accepted treatment for hemophilia.