Haemophilia

Mehroz Hasnain

1st Year MBBS, Islamabad Medical and Dental College, Islamabad, Pakistan

Key points:

- Introduction
- Risks of Haemophilia
- Clinical Diagnosis
- Problems Faced with Haemophilia
- Advancements in Treatment

Introduction

Hemophilia is a congenital genetic disorder characterized by the failure of blood to clot properly. This is an X-linked recessive disorder and has further classifications, one of which is Hemophilia C or Factor-XI Deficiency.[1]Unlike Hemophilia A (Factor VIII) and B (Factor IX), Factor XI is an autosomal disorder which means that it is equally prevalent in both males and females.[1]

Risks of Haemophilia

Despite the recent advancements in bleeding disorders and clinical diagnosis, there are still many challenges and hidden aspects that a patient suffering faces every day. For Haemophilia, the patient may bleed excessively even from a minute cut or tear, it may also cause easy bruising as well as create a higher risk of internal bleeding and permanent damage to body structures.[2]

Clinical Diagnosis

For clinical testing and diagnosis, we perform an aPTT test (activated partial thromboplastin time) that measures how long it takes for blood to clot. This is done by measuring the number of clotting factors (FXI, FVIII, FIX) present in plasma. Clinical research also shows that FXI-deficient patients have prolonged aPTTs as compared to FVIII and FIX. Severe bleeding is uncommon though, it can cause severe issues when we consider the oral cavity and urinary tract due to their rich fibrinolytic activity. This can further lead to menorrhagia epistaxis and possibly hemorrhage.[3]

Problems Faced with Haemophilia

Patients with haemophilia experience spontaneous and prolonged bleeding that may occur due to past surgery or injury. This includes bleeding episodes in joints or muscles causing swelling and restricted motion. Multiple cases reported in haemophilia suggest that damaged joint affects bone health eventually progress to chronic haemophilic arthropathy.[4]

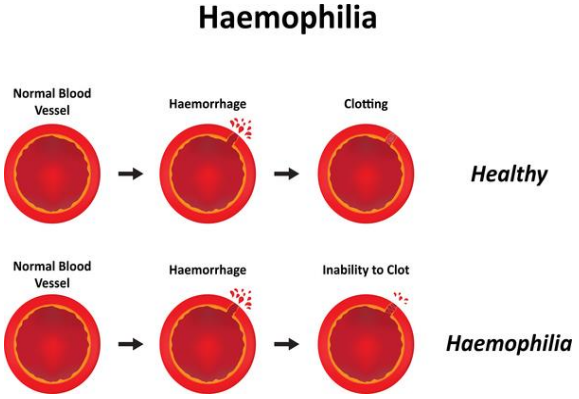

Figure 1: *Haemophilic RBC.*[6]

Advancements in Treatment

Many advancements have been made to reduce risks and mortality and improve quality of life. These include on-demand treatment, factor replacement therapy, non-replacement therapy, and surgical treatments that reduce bleeding. Furthermore, Genetic engineering has also made it possible for in vitro clotting factor synthesis, which could possibly cure patients to a certain degree.[5]

References

1. Berntorp E, Fischer K, Hart DP, Mancuso ME, Stephensen D, Shapiro AD, Blanchette V. Haemophilia. Nature reviews Disease primers. 2021 Jun 24;7(1):45
2. Nagar LK, Sharma N, Khinchi MP, Khan MS, Kumar A. HAEMOPHILIA: AN OVERVIEW. Asian J Pharm Res Dev [Internet]. 2017 Mar. 1 [cited 2025 Mar. 16];5(2):1-10
3. Wheeler AP, Gailani D. Why factor XI deficiency is a clinical concern. Expert review of hematology. 2016 Jul 2;9(7):629-37.
4. Benemei S, Mattia C, Di Minno MN. The good, the bad and the ugly of pain in haemophilia: Recent evidence on the epidemiology, molecular mechanisms and knowledge gaps preventing optimal treatment. Haemophilia. 2024 May;30(3):589-97.
5. Chowdary P, Carcao M, Kenet G, Pipe SW. Haemophilia. The Lancet. 2025 Mar 1;405(10480):736-50.
6. http://www.yogavanahill.com/diseases/classical-haemophilia-haemophilia