A2. **HEMOPHILIA: PRACTICAL CONSIDERATIONS AND CURRENT MANAGEMENT OPTIONS.**

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Hemophilia is a rare congenital bleeding disorder, resulting from a deficiency of factor VIII (hemophilia A) or factor IX (hemophilia B) which manifests with prolonged or spontaneous, potentially life-threatening bleeding. Repeated bleeding episodes, most commonly in the joints and muscles, can also result in long-term, disabling complications.

The diagnosis of hemophilia is based on factor assays, bleeding patterns, and family history.

Hemophilia can be severe, moderate, or mild, depending on the degree of factor deficiency. A common long-term complication of hemophilia is permanent damage to the joints (hemarthropathy) caused by repeated bleeding episodes. Damage to the joint tissues can be seen after even short periods of exposure to small amounts of blood. Repeated bleeding into the muscle can also have long-term effects, with muscle and nerve damage potentially leading to contractures.

Treatment of hemophilia is comprehensive and focused on preserving both physical health and quality of life of individuals with the disorder. The primary goal of treatment is the prevention or cessation of bleeding episodes. Prompt treatment of acute bleeding episodes is essential to minimize long-term complications.

Available factor VIII and IX concentrates are either plasma-derived or produced via recombinant technology. Recombinant factor products are classified as first-, second-, or third-generation based on exposure to human or animal proteins. Recombinant factor VIII products are also classified as full-length or B-domain deleted proteins. However, the 2 main concerns with factor replacement are transmission of pathogenic viruses or prions and the development of inhibitors. Factors that may increase the risk of inhibitor development include severity of hemophilia, the type of genetic mutation, family history, and age at diagnosis and first treatment.

Patient education is also key in the treatment of hemophilia, not only to ensure that patients can recognize bleeding episodes quickly, but also to allow for a higher quality of life through an understanding of the disorder and the need for comprehensive care.