Overview of Hemophilia

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What is hemophilia?

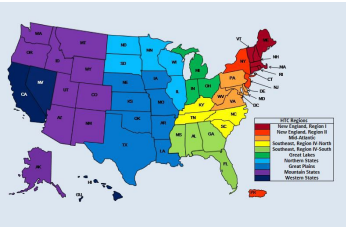
Hemophilia is a rare, complex, and potentially fatal bleeding disorder that is currently incurable and often poses physical, social and economic strain on affected individuals and their families. Bleeds are internal, and can be spontaneous. The disorder primarily affects males and is inherited in approximately 70% of cases; about 30% of cases are spontaneous mutations.[1] The condition's intensity depends upon the amount of clotting proteins, or factors, FVIII in hemophilia A (HA) and FIX in hemophilia B (HB) missing from the blood. Bleeds occur chiefly into joints and muscles; bleeds to the head, throat, abdomen or other organs can be fatal. Recurrent bleeding into joints leads to progressive cartilage and joint destruction, causing chronic pain, which increases risks for long-term opioid misuse. Persons living with hemophilia experience limited daily functioning and multiple surgeries, emergency room visits and hospitalizations. Despite treatment advances during the last 40 years, those affected still have a shorter life expectancy than the general population.[2]

HA and HB Prevalence

HA and HB both occur among all ethnic groups worldwide. However, HA is about 4 times more common than HB; prevalence at birth for hemophilia A is estimated to be 24.6 per 100,000 males and 5.0 for HB. [3]

US Hemophilia Treatment Center Network

In 1975, Congress appropriated funds to develop comprehensive care for persons with hemophilia. Today the US network of federally designated Hemophilia Treatment Centers (HTCs) includes about 150 Centers organized into eight regions (shown at left).[4] HTC network staff collect uniform data on patient demographics [5], complications [6], satisfaction,[7] health services used, and mortality. The Network's structure fosters rapid dissemination of best practices, provides training and technical assistance, and builds clinical services in underserved areas. The HTC patient population has grown and diversified, especially among traditionally underrepresented (e.g. Hispanic and African American) and under-recognized (female) populations.



The Comprehensive Care Model

HTCs use a “comprehensive care” model that provides integrated, multidisciplinary specialty care to patients of all ages and their families. The core team typically includes clinical specialists: pediatric/adult hematologists, nurse coordinator, physical therapist and social worker. Specialized laboratories are essential to ensure accurate diagnosis. HTCs provide, coordinate with, and/or refer to numerous support services, including psychologists, case managers, pharmacists, genetic counselors, orthopedists, dentists, adolescent medicine, and primary care practitioners. HTC-based comprehensive care reduces mortality by 40% among males with hemophilia, and HTC users are 50% less likely to experience a bleeding-related hospitalization. HTC patients also have higher rates of high school graduation than the general population. Many HTCs now also treat other rare inherited bleeding and clotting disorders. [8-11]

Medically Supervised Home Treatment Regimens Increase Wellness

HTCs promote “medically supervised home therapy” as standard-of-care treatment for people with severe bleeding disorders.[12] Home therapy prevents and/or decreases bleeding frequency and cumulative damage by providing rapid treatment. Empowering individuals to treat at home reduces office and emergency room visits, hospitalizations, lowers school and work absenteeism, and promotes the ability of patients and families to manage this chronic disease as independently as possible.[13] HTCs provide extensive education to ensure that use of home therapy is implemented appropriately. In affluent countries, hemophilia care employs one of two medication regimens: prophylactic or episodic treatment.

Prophylaxis Treatment Regimens

Prophylactic treatment involves the regularly scheduled administration of medications to prevent bleeding. The overall goal is to avoid joint damage and disability. Prophylaxis allows most people with hemophilia to lead healthy and active lives. Prophylaxis regimens typically require intravenous infusions, two to four times per week, depending on hemophilia severity, bleeding phenotype, and pharmacokinetic characteristics. During the last decade, prophylactic regimens tailored to the individual needs of patients using pharmacokinetic modeling have become more widely used. Tailored regimens can reduce factor consumption and facilitate more cost-effective use of expensive medical resources. [14]

Episodic Treatment Regimens

In episodic treatment regimens (also known as on-demand therapy), medication is administered after a bleed starts. Episodic treatment reduces pain and the debilitating impact of individual bleeds. However, it cannot

alter the overall progressive deterioration that leads to musculoskeletal damage and other complications. Episodic treatment remains useful for people who experience bleeding episodes annually or less often.

Novel Therapies

Two main types of new therapeutic products for both HA and HB that work longer than conventional hemophilia medications, have become available over the last five years. The first type, known as "extended half-life" (EHL) products, require fewer intravenous infusions than the legacy clotting factor concentrates (CFCs). The second new type is a non-CFC treatment which is administered under the skin ("subcutaneously") rather than intravenously. Non-CFC treatments can be administered once weekly, or 1-2 times per month. Because these novel therapies work longer between doses, individuals with hemophilia and their families can now participate in more activities than ever, without fear of bleeding. [15]

Gene Therapy

Gene therapy (GT) holds promise for a lasting cure with a single drug administration. GT uses a modified virus to insert a functional copy of the absent or nonfunctional gene into the body, stimulating production of the missing protein needed for normal blood clotting. [16] One gene therapy product has been approved by the FDA as of October 2023. However, much research remains before gene therapy can be determined safe and effective.

Policy Considerations

People with Hemophilia are at high risk for frequent serious medical disruptions that can impede full physical and social functioning and economic productivity. Strategic health policy can reduce these risks by promoting interventions that optimize health. Policies should include these elements: 1) access to the team-based regional model of HTC specialty diagnostic, treatment, and outpatient pharmacy services to address patient/family physical and psycho-social needs and integrate clinical pharmacy into HTC care; 2) optimal reimbursement for both the comprehensive team-based outpatient care model to ensure its sustainability, and for new therapies to provide effective, safe, affordable and sustainable treatment; and 3) workforce development via loan repayment to incentivize healthcare professionals to commit to careers in rare hematologic disorders.

References

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