



## Hemophilia

Hemophilia is an inherited bleeding disorder that results from a low level or lack of proteins needed for normal blood clotting. People with hemophilia have a predisposition towards spontaneous internal bleeding and prolonged bleeding after an injury or surgery. Over time, these bleeding episodes can cause severe damage to joints, nerves, and other body systems and, in rare cases, may cause death.

Because the genetic mutation that causes hemophilia is located on the X chromosome, males are the most affected, while females are most likely to act as carriers and can have a mild form of hemophilia. Hemophilia affects 1 in 5,000 male births. About 400 male infants are born with hemophilia each year. Over their lifespan about 15-20% of people with hemophilia will develop an antibody (inhibitor) to the treatment product which decreases the effectiveness of the product to stop bleeding.

The National Center on Birth Defects and Developmental Disabilities' Division of Blood Disorders is committed to preventing and controlling complications resulting from bleeding disorders, such as hemophilia.

### 2012 Accomplishments

- Created a database of more than 2,000 Factor VIII (8) gene mutations reported to cause hemophilia worldwide. The mutation that a person has determines how much clotting factor is produced and how well it works. Knowing the mutation that a person with hemophilia has is important for genetic testing of family members. It also can help in predicting how likely a person is to develop an inhibitor. To view the database, visit: [cdc.gov/hemophiliamutations](http://cdc.gov/hemophiliamutations).
- Produced two videos for youth with hemophilia: (1) *Playing it Safe with Hemophilia*, and (2) *Starting the Conversation: How to talk to your friends about hemophilia*. The videos were developed using the findings from formative research that included testing messages and video prototypes with focus groups comprised of youth with hemophilia. To view the videos, visit: [cdc.gov/ncbddd/socialmedia/videos.html](http://cdc.gov/ncbddd/socialmedia/videos.html)
- Examined the average annual health care expenditures for people with hemophilia covered by employer-sponsored insurance. Data from the MarketScan Commercial and Medicare Research Databases were used for the period 2002-2008 to identify cases of hemophilia and to estimate mean and median medical expenditures during 2008.

## **2012 Accomplishments (continued)**

- Updated and improved the bleeding disorders surveillance system known as the Universal Data Collection (UDC) surveillance project to collect additional information on emergent issues, treatment, complications, and the use of health care services. The UDC was initially implemented in the late 1990s in response to HIV/AIDS transmission from blood products used to treat people with hemophilia. Today, the population of people with bleeding disorders is more diverse and susceptible to different complications, including emerging pathogens, chronic diseases, and inhibitor development.
- The American Thrombosis and Hemostasis Network in partnership with the Hemophilia Treatment Center (HTC) Network were funded to collect information from people with hemophilia and other bleeding disorders who receive care at HTCs. Duke University Medical Center and the Hemophilia Federation of America are collecting information from people with bleeding disorders who receive care elsewhere to better capture the health needs of the hemophilia population.
- Hosted a meeting of partners in hemophilia treatment, industry, and government to review data and discuss implementation issues relevant to planned national inhibitor surveillance by CDC in collaboration with the federally funded HTC Network. Inhibitors, or antibodies to the treatment product used to prevent and treat bleeding in people with hemophilia, are a serious complication of hemophilia care. The burden of this complication in the hemophilia population is not known, therefore surveillance is needed.
- Hosted the 2nd National Conference on Blood Disorders in Public Health. The purpose of this conference was to promote the health and improve outcomes among people with blood disorders.

### **Jane's Story**



Jane is the mother of Leland, a young man with hemophilia. To read Jane's story, visit: [www.cdc.gov/ncbddd/hemophilia/stories.html](http://www.cdc.gov/ncbddd/hemophilia/stories.html)

## Looking to the Future

There is still much to learn about bleeding disorders such as hemophilia, and how to best prevent and manage complications associated with them. As people with hemophilia live longer lives than those in previous generations, they face new challenges related to chronic age related health problems such as heart disease, high blood pressure and diabetes. The causes and risk factors for developing inhibitors are poorly understood. However, the specific genetic mutation causing hemophilia for an individual may help predict what treatments will decrease the risk for developing an inhibitor.

Strengthening CDC's bleeding disorder programs and activities can fill the public health needs essential for improving the quality of life for people with hemophilia. The National Center on Birth Defects and Developmental Disabilities' Division of Blood Disorders is committed to collecting and analyzing data, conducting epidemiologic research, enhancing laboratory capacity, and supporting hemophilia education and outreach activities through our community-based partners. We are striving to reduce the morbidity and mortality in people with hemophilia through comprehensive public health practice by contributing to a better understanding of hemophilia and its complications; ensuring that prevention programs are developed, implemented, and evaluated; developing information that is accessible to consumers and health care providers; and implementing programs that can improve the quality of life for people living with hemophilia.

## Notable Scientific Publications

Guh S, Grosse SD, McAlister S, Kessler CM, Soucie JM. Healthcare expenditures for males with haemophilia and employer-sponsored insurance in the United States, 2008. *Haemophilia* 2012; 18:268-75.

Guh S, Grosse SD, McAlister S, Kessler CM, Soucie JM. Health care expenditures for Medicaid-covered males with haemophilia in the United States, 2008. *Haemophilia*. 2012; 18:276-83.

Miller CH, Platt SJ, Rice AS, Kelly F, Soucie JM; the Hemophilia Inhibitor Research Study Investigators. Validation of Nijmegen-Bethesda Assay modifications to allow inhibitor measurement during replacement therapy and facilitate inhibitor surveillance. *J Thromb Haemost* 2012; 10:1055-61.

### Did You Know?

- Children who were treated on a regular basis to prevent bleeding had less evidence of joint damage.
- About 15% to 20% of people with hemophilia develop an inhibitor that results in the body's resistance to treatment products used to treat their bleeding disorder. A single patient with complications and an inhibitor can have over \$1M in treatment costs annually.
- People with hemophilia who develop an inhibitor are twice as likely to be hospitalized for a bleeding complication.

**Hemophilia**  
**Annual Report, Fiscal Year 2012**

**Notable Scientific Publications** (continued)

Payne AB, Bean CJ, Hooper WC, Miller CH. Utility of Multiplex Ligation-Dependent Probe Amplification (MLPA) for Hemophilia Mutation Screening. *Journal of Thrombosis and Haemostasis* 2012;10(9):1951-1954.

Payne AB, Miller CH, Kelly FM, Soucie JM, Hooper WC. The CDC hemophilia A mutation project (CHAMP) mutation list: a new online resource. *Human Mutation* 2012;epub: Dec 26

Soucie JM, De Staercke C, Monahan PE, et al. Evidence for the Transmission of Parvovirus B19 in Patients with Bleeding Disorders Treated with Plasma-derived Factor Concentrates in the Era of Nucleic Acid Test (NAT) Screening. *Transfusion* 2012, Sept 24 epub before print.

Carpenter SL, Soucie JM, Sterner S, Presley R; Hemophilia Treatment Center Network (HTCN) Investigators. Increased prevalence of inhibitors in Hispanic patients with severe haemophilia A enrolled in the Universal Data Collection database. *Haemophilia*. 2012; 18:e260-5.

Kempton CL, Allen G, Hord J, Kruse-Jarres R, Pruthi RK, Walsh C, Young G, Soucie JM. Eradication of factor VIII inhibitors in patients with mild and moderate hemophilia A. *Am J Hematol* 2012; 87:933-936.

To view the annual report online, visit:  
**[www.cdc.gov/ncbddd/2012AnnualReport](http://www.cdc.gov/ncbddd/2012AnnualReport)**

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