Indiana

Hemophilia &

Thrombosis

Center, Inc.

Hemophilia Awareness Month – Fast Facts for Physicians

Hemophilia is an inherited, X-linked, recessive disorder caused by a deficiency or absence of functional factor VIII, IX or XI, (hemophilia A, B and C, respectively). In the United States, approximately 400 infants are born each year with the condition. To aid in the understanding of hemophilia, the most common myths about the disorder are addressed below.

Myth 1: Patients who have hemophilia have a known family history of the disorder.

- Spontaneous genetic mutations are responsible for 30% of new cases, and, therefore, hemophilia may occur in families who do not have a history of the disorder.
- Due to the absence of significant bleeding episodes, often as a result of having a mild to moderate deficiency, some families are not aware of a history of hemophilia in their family.
- Women may be unaware of their carrier status, as they have passed on the disease-causing gene unknowingly to females for generations before an affected male receives the gene and presents it clinically.

Myth 2: Individuals who have hemophilia are diagnosed at birth.

- Many people who have hemophilia are not diagnosed at birth, either due to the absence of a family history or clinical bleeding symptoms.
- Although people who have severe hemophilia are usually diagnosed early in life, those who have a mild or moderate deficiency may not be diagnosed until they sustain an injury or undergo an invasive procedure that precipitates a bleeding episode.
- Acquired hemophilia is a rare condition caused by the development of alloantibodies directed against one of the coagulation factors, typically factor VIII. This condition is not inherited, and previously normal individuals may therefore develop hemophilia.

Myth 3: Hemophilia primarily affects Caucasians.

- Hemophilia occurs in all races and ethnic groups equally.
- In the United States, hemophilia rates are similar in Caucasian, African American, Asian and Hispanic males.

Myth 4: Only males have hemophilia.

- Although instances are rare, females can have hemophilia. This occurs if the female co-inherits the hemophilia gene from her father and the carrier gene from her mother. It may also occur in disorders in which only one X chromosome is present (such as Turner's syndrome).
- Lyonization imbalance can result in carriers with low levels of clotting factor protein, which usually places them in the mild range of deficiency. In some rare circumstances, carriers may have moderate or severe disease.
- There are other rare bleeding disorders that affect males and females equally, including hemophilia C (Factor XI deficiency), type 3 von Willebrand disease and deficiencies of factors VII, X, V etc.

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Myth 5: Individuals with hemophilia bleed faster and may experience severe bleeding due to a superficial cut.

- People with hemophilia bleed longer, not faster, due to their inability to form a stable fibrin clot.
- Typically, minor bleeding—especially from superficial cuts—is neither life threatening nor excessive. Important exceptions do, however, exist, such as bleeding in the oral cavity, where it is often difficult to gauge the amount of blood loss due to swallowing and saliva.

Myth 6: People with hemophilia have a shortened life-span and are usually disabled.

- Most people who have hemophilia now lead long, full and productive lives.
- Although the average lifespan of a person with severe hemophilia in the 1960's was 11 years, the advent of home replacement therapy and viral inactivation of clotting factor concentrate allows those born with hemophilia today to have a normal life-expectancy.
- Since the initiation of prophylaxis in the mid-1990's, children who have hemophilia lead a life of less pain and disability, primarily due to the prevention of hemorrhagic episodes and resultant orthopedic damage.

Myth 7: Physical activity is not recommended for people with hemophilia.

- Regular exercise strengthens muscles and joints, ultimately resulting in injury prevention.
- Exercise is very important in weight management and is encouraged for patients who have hemophilia. Overweight patients with hemophilia often have decreased range of motion and increased morbidity due to their weight.
- A list of sports that are encouraged for people who have hemophilia can be found in *Playing it Safe: Bleeding Disorders, Sports and Exercise,* a National Hemophilia Foundation publication.

Myth 8: Everyone with hemophilia is infected with a viral disease, such as HIV and/or hepatitis C, transmitted by blood or blood derivatives.

- Unfortunately, in the 1980's many patients that used clotting factor concentrate for treatment of hemophilia acquired HIV (60-90% of those with hemophilia A; 30-50% of those with hemophilia B) and/or hepatitis C (approximately 80% of all people with hemophilia) that was present in the blood supply.
- Today, improved donor screening, advanced viral inactivation techniques and purification methods for plasma-derived products has made the treatment of hemophilia very safe. In addition, there are now genetically engineered products available for treatment of hemophilia A and B.
- There has not been a documented case of HIV or hepatitis C infection transmitted from clotting factor concentrate use since 1987.
- The majority of hemophilia patients today *are not* infected with HIV or hepatitis C.

Myth 9: The aPTT is always prolonged in someone with hemophilia.

- The aPTT may not be prolonged in those who have mild or moderate factor VIII or IX deficiency and is therefore not always a reliable screening test.

Myth 10: An intracranial hemorrhage occurs after a head injury and always occurs with visible signs.

- An intracranial hemorrhage may occur without loss of consciousness, visible bruises or a hematoma.
- Symptoms may take time to emerge, and report of a head injury may not always be present.
- As intracranial hemorrhage is an important cause of morbidity and mortality in those who have hemophilia, aggressive evaluation and awareness is crucial to early detection and treatment.

Myth 11: All physicians and medical facilities are prepared and equipped to provide optimum care for hemophilia patients.

- Care from all providers is *not* equal.
- The Centers for Disease Control and Prevention has documented that persons who receive care in a hemophilia treatment center (HTC) have a 40% lower risk of mortality (p=0.003), compared to those seen outside the network.
- Those seen within an HTC also have a substantially lower risk of bleeding complications. Furthermore, patients seen within the network are 40% less likely to be hospitalized for complications associated with bleeding.[1]
- The Indiana Hemophilia and Thrombosis Center (IHTC) is the ***only*** federally recognized HTC in Indiana.

More information about hemophilia care and treatment is available through the National Hemophilia Foundation (1-800-424-2634) or from Hemophilia of Indiana (1-317-396-0065).Visit the Indiana Hemophilia and Thrombosis Center (IHTC) website (ihtc.org) for detailed information about the center, including the comprehensive care team, educational newsletters, pharmacy and specialty clinics

References:

1.Soucie JM, Symons J, Evatt BL, et al. Home-based factor infusion therapy and hospitalization for bleeding complications among males with haemophilia. Haemophilia 2001;7:198-206.