Your Guide to Mild Hemophilia
What is Hemophilia?

- Hemophilia is a bleeding disorder that prevents blood from clotting normally.
- Hemophilia is caused by a shortage or deficiency of one of the proteins in blood responsible for blood clotting.
- There are two major types of hemophilia—Hemophilia A or a deficiency of the factor VIII protein; and Hemophilia B, a deficiency of the factor IX protein.
- Hemophilia is a genetic disorder that may be “carried” by females, but usually only males actually have symptoms of hemophilia.
- Female hemophilia carriers with lower than normal levels of factor VIII or IX may have bleeding problems similar to males with mild hemophilia. Some female carriers have normal clotting protein levels and do not have abnormal bleeding.
- Hemophilia occurs in varying degrees of severity depending upon the amount of normal clotting protein that the individual can manufacture. Individuals with very small amounts of factor VIII or factor IX have the most severe symptoms. Those with nearly normal amounts (mild hemophilia) usually have very mild symptoms.
- A person with mild hemophilia will usually not bleed unless he sustains a major injury or has surgery or a dental procedure.
Both type of hemophilia and severity are inherited characteristics and will be the same in all affected family members.

There is no “cure” for hemophilia at the present time but there are treatments to prevent or to stop bleeding and prevent complications of the disease.

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<th>Factor VIII or IX level in blood</th>
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Diagnosing Mild Hemophilia

- There are two ways to diagnose hemophilia – by taking a blood sample to determine factor VIII or IX activity level, or if there is a known family member with mild hemophilia, DNA testing can be done to confirm the diagnosis in additional family members.
- DNA testing may also be done prenatally when a female carrier and her family would like to know if their unborn child will have hemophilia.
- An infant may be diagnosed shortly after birth, either by testing blood from the umbilical cord or by drawing blood from the baby’s vein.
- If there is no family history of hemophilia, the diagnosis is often not made during the childhood years unless the child has had some significant trauma or a surgical procedure.

These might include:

- Circumcision
- Tonsillectomy
- Dental extractions
- Sports injuries
- Accidents

- Diagnosis in teens and adults can occur following unexplained bleeding during or after a medical procedure, surgery, or sutures, or following a sports injury or other injury that required a longer than normal time for healing.
- Many adult males are diagnosed when surgery is scheduled and pre-operative screening tests indicate that they may be at risk for excessive bleeding. A referral for additional laboratory studies often reveals a diagnosis of mild hemophilia.
• Some males are diagnosed when a common surgical procedure (such as appendectomy, prostate surgery, hernia repair, gall bladder surgery or dental extraction) is complicated by excessive bleeding or prolonged healing.

• A primary care physician or a hematologist may diagnose hemophilia but referral to a hemophilia treatment center (HTC) should take place soon after diagnosis.

• Once diagnosis is confirmed it is important to have a family “pedigree” constructed so that family members who may be at risk of having hemophilia or of being a hemophilia carrier may be alerted. At risk individuals can then be offered laboratory testing and counseling.

• Correct diagnosis and education are always the best ways to prevent bleeding complications from occurring. Education is most effective when done at a scheduled appointment in a calm environment that is conducive to open discussion regarding diagnosis and the development of a hemophilia treatment plan. The newly diagnosed individual should have ample opportunity to have any and all questions regarding hemophilia diagnosis and management answered by knowledgeable hemophilia experts.

Genetics of Hemophilia

• Hemophilia is a sex-linked (or X-linked) genetic disorder. It is carried by females, but only males actually have hemophilia with its bleeding complications.

• The gene for hemophilia is carried on the X chromosome
• Females have two X chromosomes. In hemophilia, one of the X chromosomes is normal and one of them carries the hemophilia gene. The hemophilia X chromosome results in a deficiency or absence of the factor VIII or IX protein required for normal blood clotting.

• In a female carrier of hemophilia, the normal gene overrides the hemophilia gene and although the female carries the disorder, she does not fully express the disease and usually does not have major bleeding symptoms.

• Males have an X and Y chromosome. The Y is inherited from their father and the X is inherited from their mother. If a male child inherits the mother’s hemophilia X, he will be affected by hemophilia. If he inherits her normal X, he will not have hemophilia.

• Each son of a hemophilia carrier has a 50% chance of having hemophilia; each daughter has a 50% chance of being a carrier. Carrier status can be accurately determined by having blood drawn for DNA testing.
Hemophilia Carriers

• The first indication of whether someone may be at risk for being a carrier is the family history. If there are affected males in a family, then some females are at risk of being carriers.

• Women that have at least two sons with hemophilia, or one son and another blood relative with hemophilia are proven hemophilia carriers.

• Women that have one affected son or no other blood relatives with hemophilia may or may not be carriers. They will need genetic testing to determine whether they carry the gene for hemophilia.

• Obligate carriers are the daughters of men with hemophilia. They must inherit the hemophilia X from their father and a normal X from their mother. This XX combination automatically makes them obligatory carriers.

• Obligate carriers do not need genetic testing to determine whether they carry the hemophilia gene. They do and they have a 50% chance of passing the gene to each of their children.

• In 1 out of 3 families where hemophilia has never been diagnosed before, a new and spontaneous mutation may have occurred which caused hemophilia to occur in a family member.

• Both carriers and obligate carriers need to have factor VIII or IX activity levels determined by a laboratory analysis. Many carriers have lower than normal factor levels. These levels can be as low as those in mild hemophilia and may put the carrier at risk for bleeding.
• If a carrier has low (50% or less) factor VIII or IX levels, she may need clotting factor or DDAVP for injuries, dental or surgical procedures. She should contact the Hemophilia Treatment Center if any procedures are being planned so that a treatment plan can be formulated.
• In an emergency situation, when a carrier’s factor VIII or IX level has not yet been determined, it should be assumed that her level is <50% and appropriate treatment should be administered.

**Pregnancy**

➢ When pregnant with a male child, some carriers may elect to have prenatal DNA testing done to determine if the fetus is affected by hemophilia. 
➢ There are several tests available including: ultrasound (for sex determination), chorionic villus sampling (CVS), amniocentesis, and percutaneous umbilical blood sampling (PUBS). With the exception of the ultrasound, there are some risks associated with the other tests that need to be discussed with the obstetrician or geneticist. Factor activity testing may be performed prior to the procedure and in some cases treatment may be required for the pregnant woman. 
➢ If a pregnant carrier does not wish to have prenatal testing, a treatment plan for the pregnancy, delivery and post-partum period should be formulated that assumes the male child is affected by hemophilia. This will allow precautions to be taken that will insure the safety of an infant with a potential bleeding disorder. 
➢ All pregnant hemophilia carriers should be seen at the Hemophilia Treatment Center for updated information and specialized care. HTC staff will work with the obstetrician/gynecologist to plan appropriate care.
Treatment

Bleeding
➢ Bleeding episodes respond best if treated promptly by raising the clotting factor at least above the 50% level to stop the bleeding
➢ If in doubt as to whether treatment is needed, contact your HTC or hematologist to discuss your injury or planned surgery

General Care
➢ Clean superficial wounds with soap and water, apply pressure until bleeding stops, and use a Band-aid or a pressure bandage if necessary
➢ More serious lacerations in the person with hemophilia require the same first aid treatment as a person without hemophilia, however if sutures are needed to close a wound, it is important to seek treatment recommendations from a physician at the HTC or a hematologist with hemophilia expertise.
➢ For muscle, joint or soft tissue injuries or trauma the first line of treatment is RICE

Rest – rest the injured/bleeding part which may include a sling or crutches
Ice – apply ice over a thin cloth; flexible ice pack or bagged loose ice is best. Leave the ice pack on for no longer than 20 minutes at a time. This should be done several times a day for the first 48 hours following an injury
Compression – use carefully applied compression bandage where appropriate.
Elevation – raise the affected area above the level of the heart

➢ While each patient and each bleeding episode may be handled differently, the following factor concentrates or medications are used in the treatment of mild hemophilia
Treatment Products

Hemophilia A

Desmopressin Acetate (DDAVP):
- DDAVP may be the treatment of choice for some people with mild hemophilia A. This synthetic hormone can be administered either intravenously (in the vein), subcutaneously (under the skin) or as a nasal spray called Stimate® (1.5mg/ml strength).
- DDAVP works by causing Factor VIII stored in the lining of blood vessels to be released into the blood stream to control bleeding. While response to DDAVP varies among individuals and even family members, it can raise one’s level by 3-fold (3 times the circulating level or baseline level of factor VIII).
- No individual’s response to DDAVP is predictable, thus making it important to have a test dose with levels of Factor VIII measured before and after the dose. This will help the HTC determine if and when this drug can be used to control bleeding.
- Some people do not respond to DDAVP or may have other health conditions that would prevent the safe use of this medication thus eliminating DDAVP as a treatment option for that person. **DDAVP is not effective for Factor IX deficiency**
- While DDAVP (or Stimate®) is generally safe for most people, there are potential side effects and precautions to be considered. If a person requires more than two doses of DDAVP or Stimate for the same bleeding episode, the storage sites might be temporarily depleted causing inadequate factor VIII to be released. Depending upon the situation, a clotting factor concentrate might be required.

Factor VIII Concentrates:
- There are numerous factor VIII products available ranging from plasma-derived products to products...
that are primarily recombinant with plasma protein additions to completely synthetic factor made in the laboratory.

- All three types are currently available for patient choice. All of these concentrates come as a freeze-dried powder which is mixed with sterile water and administered into a vein.
- The Medical & Scientific Advisory Council (MASAC) of the National Hemophilia Foundation states that recombinant factor VIII products are the recommended treatment of choice for patients with hemophilia A. (MASAC Guideline #151).
- While there is a theoretical risk of viral or other pathogenic transmission from the use of clotting factor concentrate, it is worth noting that there have been no reports of pathogens being transmitted by any of these products in 20 years.

**Hemophilia B**

**Factor IX Concentrates:**

- Persons with a deficiency of factor IX have hemophilia B and require a factor IX clotting concentrate to prevent or treat bleeding
- As with factor VIII products, factor IX concentrates can be either plasma derived or recombinant (genetically engineered)
  - Prothrombin Complex Concentrates. Plasma derived factor IX products that also contain other clotting factors and have the potential to cause excessive clotting
  - Highly purified plasma derived factor IX concentrate contains only factor IX.
  - Recombinant factor IX concentrate contains no human or animal proteins
- All classifications of factor IX products are available for patient choice
Preventing Bleeding

As a person with mild hemophilia there are things you can do to help prevent bleeding:

*Always notify your HTC if you are planning to have an invasive procedure, surgery or dental work so that treatment recommendations can be made to prevent bleeding.*

**These procedures might include:**

**Dental work**
- Routine cleaning
- Fillings requiring injection for anesthesia
- Dental procedures such as tooth extraction, or work on gum tissue

**Surgical procedures**
- Invasive or diagnostic procedures
- Eye surgeries
- Colonoscopy
- Biopsies

**Immunizations**
- Subcutaneous or intramuscular
When an injury occurs remember first and foremost you have a bleeding disorder. Contact your HTC for recommendations for treatment if you have an injury.

Watch for signs and symptoms of bleeding
- Swelling of a joint, soft tissue or muscle
- Pain
- Area is warm to touch

Injury or blow to the head, neck or abdomen
- Notify your HTC immediately for evaluation and treatment

Things to avoid:
- Contact sports such as football or hockey; as well as other high risk activities
- Aspirin or aspirin containing products

Be prepared when traveling
- Contact your HTC before traveling
- Always carry your factor product, supplies or Stimate® with you
- Carry a letter explaining your hemophilia, need for treatment, dosage and HTC contact information
- Obtain contact information for the HTC in the area to which you are traveling in order to access appropriate hemophilia care if needed
Be Proactive

Wear protective gear such as helmets and knee pads when doing activities that may put you at risk for injury and bleeding.

Practice good dental hygiene to avoid tooth decay and gum disease.

Eat a nutritional diet and maintain a healthy weight.

Exercise regularly for strong muscles and healthy joints.

Wear a medic alert bracelet or necklace.

Get immunized against Hepatitis A and Hepatitis B.

Notify all of your physicians including your dentist about your hemophilia.

Use caution with homeopathic or herbal remedies.

Get yearly comprehensive hemophilia evaluations.

Always maintain health insurance.
Conclusions

● Mild hemophilia is a very manageable disorder when appropriate treatment is sought.

● A knowledgeable patient is a safe patient—seek information and education regarding hemophilia.

● Regular contact with the HTC is highly recommended even when there are no obvious bleeding problems. Once a year or an every other year visit is optimal for good preventive management.

● Contact your HTC or hematologist prior to any invasive procedures, surgery or dental work for appropriate treatment recommendations.

● Remember that an INJURY EQUALS A BLEEDING EPISODE. Don’t just treat the injury. Call the HTC and inquire about your bleeding risk.

● Live life to the fullest and have fun!!
My Hemophilia Diagnosis:

Type of Hemophilia

Severity