

1 HEMOPHILIA TYPE A

2 Hemophilia A is...

☞ a blood clotting disorder caused by a mutation of the factor VIII (the body's essential clotting factor), It is the most common hemophilia. Inheritance is linked to the X chromosome and 1 in 10,000 males are affected, and very few females.

3 History Of Hemophilia

- Hemophilia was recognized, though not named, in ancient times, around 2nd century AD.
- Hemophilia has often been called "The Royal Disease" due to its occurrence in the European royal families.

4 History Of Hemophilia Part 2

5 Causes of Hemophilia A

- It is caused by a mutation of a the Factor VIII gene
- It is passed on from parent to child (primarily mother to son though a father can pass it to a son as well, or in very rare instances both parents can pass it on to a daughter).

6 Getting Hemophilia A

- Male children are born with hemophilia A by receiving a mutated X chromosome from the mother (if the mother has a mutated gene on one of her X chromosomes the chances of her son having hemophilia A are still 50/50.

7 Getting Hemophilia A part 2

- For a female child to inherit the disease her mother must carry (or have) the disease and her father must have it. Even if the mother is a carrier the chances of the daughter having it are still 50/50.

8 Symptoms of Hemophilia A

Hemophilia leads to a severely increased risk of bleeding from common injuries. The sites of bleeding are:

- ☞ joints
- ☞ muscles
- ☞ digestive tract
- ☞ brain

☞ The muscle and joint hemorrhages are quite typical of hemophilia

9 Testing for Hemophilia A

Many blood clotting tests are performed if the person tested is the first one in the family to have a bleeding disorder. Once the defect has been identified, other family members will need less testing to diagnose the disorder.

- Normal Prothrombin time (PT) and Partial thromboplastin time (PTT) (these are tests that measures the clotting time of plasma in the liquid portion of blood)
- Normal bleeding time (this is a test that measures the speed at which small blood vessels close off to stop bleeding and platelet function)
- Normal fibrinogen level (this test measures the amount of fibrinogen in the blood)
- Low serum factor VIII activity (measures the activity of factor VIII)

10 Diagnosis

- Generally, the first bleeding episode occurs before 18 months of age, often after a minor injury. Usually at this point the child with hemophilia is diagnosed.

11 Cures for Hemophilia A

All though there is no cure for hemophilia there are three main types of treatment for it:

- Prophylaxis
- Desmopressin
- On-Demand

12 Prophylaxis

- Prophylaxis is the injection of a replacement Factor VIII protein into a hemophiliac. The replacement is nearly identical to the protein which is lacking in the blood of hemophiliacs. After an infusion of the concentrate, all the proteins needed for clotting are in place. A hemophiliac's blood becomes "normal", at least for a few hours. This allows the time for a clot to form at the site of the damaged blood vessel.

13 Desmopressin

- Desmopressin is a synthetic drug that is a copy of a natural hormone. It is useful in treating people with mild or moderate hemophilia A. But unfortunately is not a treatment for people with severe hemophilia A.

14 Ways Desmopressin Can be Taken:

- It can be injected into a vein. Most often, the brand name for this kind of desmopressin is DDAVP.
- It can be injected under the skin. The brand name for this kind of desmopressin is often Octostim.
- It can be taken by nasal spray. The brand name of the nasal spray is often Octostim Spray

15 On-Demand

- On-demand therapy is the infusion of factor concentrates immediately after

the beginning of a bleed. The goal is to stop the bleeding quickly, before any damage is done to the joint or muscle

16 Organizations

- COTT: The Committee of Ten Thousand
- The Hemophilia Federation
- HANY: Hemophilia Association of New York, Inc.
- NHF: The National Hemophilia Foundation
- NCHF: Northern California Hemophilia Foundation
- Hemophilia Foundation of Oregon
- America Online (AOL) Hemophilia Group

17 Conclusion

- Hemophilia is a genetic disease that prevents blood from clotting normally
- It is caused by a mutation on the X chromosome
- Complications/loss of blood can be fatal
- Though it can not be cured it can be treated