NATIONAL HEMOPHILIA PROGRAM
COORDINATING CENTER

Before and After Genetic Testing

athn™
american thrombosis
& hemostasis network

Securing Data. Advancing Knowledge. Transforming Care.

You’ve made an important decision.

Now, find out what you need to know before and after your genetic testing for an inherited bleeding disorder.

BEFORE GENETIC TESTING

Do I need genetic testing if I already have a known diagnosis of hemophilia? No.

➜ A clear diagnosis of hemophilia can usually be made without genetic testing.

Will genetic test results confirm my diagnosis of hemophilia? Yes.

➜ Genetic testing will confirm a diagnosis of hemophilia.

➜ In some cases, testing may help make a diagnosis of mild hemophilia.

➜ It is the best way to determine if someone is a carrier of hemophilia.

Will genetic testing help define the severity of my hemophilia? Maybe.

➜ An individual's disease severity is based on the level of certain proteins in the blood (this is his or her factor level).

➜ Some gene changes (or mutations) that cause hemophilia always result in severe hemophilia.

➜ Other genes may be associated with how often one bleeds and may help explain why family members with the same diagnosis have different bleeding patterns.

Will genetic testing help predict if I will develop an inhibitor? Maybe.

➜ Studies show that inhibitors are more likely to develop in people with certain mutations (gene changes).

➜ Genetic testing may help predict your chances of developing an inhibitor. However, the mutation is only one part of why an inhibitor develops.

➜ Other factors such as race and ethnicity may also play a role.

How will I react to my genetic test results?

➜ People react differently to learning about their genes.

➜ You might want to think about how this information may make you feel.

➜ Consider having a friend or family member with you when you hear your results.

BEFORE GENETIC TESTING

Will my genetic test results help my family members? Yes.

→ Your family members who have hemophilia or are carriers will usually have the same mutation as you do.

→ Genetic testing is the most accurate way to identify women and girls in your family who carry the gene for hemophilia.

→ When the family mutation is known, carrier testing is better and costs less.

→ Your provider can help you figure out who in your family may benefit from your test results.

Should I be concerned about genetic discrimination based on genetic test results? No.

→ The Genetic Information Nondiscrimination Act of 2008 (GINA) makes it illegal for health insurers or employers to discriminate against most individuals based on genetic information.

→ Genetic testing for hemophilia is unlikely to affect your job or health insurance.

→ However, genetic information can legally be used by insurers for decisions about other types of insurance, such as life, disability and long-term-care insurance. Individuals may experience discrimination for reasons other than genetic test results.

→ If you're worried about discrimination based on genetic testing or any other factor, you're encouraged to talk to your provider.

What type of results might I get back from genetic testing?

→ Positive: The lab found a mutation that causes hemophilia.

→ Negative: The lab did not find a mutation that causes hemophilia.

→ Variant of unknown significance: The lab found a mutation, but it is unclear what that mutation means.

→ Genetic testing may show that someone is not a blood relative.

AFTER GENETIC TESTING

I have my results. Now what?

- ➔ Ask questions until you understand the results.
- ➔ Consider sharing results with other family members.
- ➔ Keep a copy of your results. You can give copies to other people.

What feelings will I have after receiving genetic testing?

- ➔ Feelings vary from person to person. It's normal to feel happy, sad, angry or guilty.
- ➔ If you're uncomfortable, you should talk with your provider about your feelings.

Will the results affect my treatment?

- ➔ Today, treatment for hemophilia is not based on an individual's mutation.
- ➔ In the future, your mutation may impact your care.
- ➔ Some specific mutations may help to predict risk of developing an inhibitor.

Who in my family should I share my results with?

- ➔ Consider sharing your results with a supportive family member or friend.
- ➔ Your provider can help identify who might benefit from knowing the results.
- ➔ Sharing your test results may help family members who choose to have genetic testing.

What should I share with family members?

- ➔ A copy of your test report stating the family mutation.
- ➔ The type of hemophilia in the family.
- ➔ The severity of hemophilia in the family.
- ➔ The family history of hemophilia (family tree). This should show who has hemophilia and who is a known carrier.

For more information about hemophilia and genetic testing, contact your local hemophilia treatment center (HTC). A national list of HTCs can be found in the Affiliate Directory under the Affiliates tab at athn.org.

To find a genetic counselor in your area, visit nsgc.org.

About the National Hemophilia Program Coordinating Center

The American Thrombosis and Hemostasis Network (ATHN) is a non-profit organization committed to improving care for individuals affected by bleeding and clotting disorders. In June 2012, the Maternal and Child Health Bureau of the Health Resources and Services Administration funded ATHN to establish the National Hemophilia Program Coordinating Center. The NHPCC serves as a bridge between the regional HTC networks, helping to create value on a national scale. The NHPCC partners with regional leadership, over 135 ATHN-affiliated HTCs, patient advocacy groups, government partners, payers, and thought leaders to guarantee a community-wide perspective. **To learn more about the NHPCC, visit athn.org.**

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