HARP
Hemophilia A
Research Program

Hemophilia A Genetic Testing

What is Hemophilia A?

Hemophilia A is a **genetic bleeding disorder** caused by low levels of **factor VIII**, a protein needed for normal blood clotting.

When factor VIII levels are low, clots are fragile, and bleeding is harder to control.

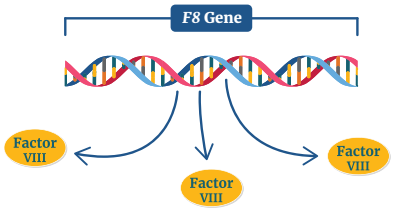
What is DNA?

DNA is the molecule inside cells that contains instructions for our bodies. DNA is made of strings of chemical letters called nucleotides: A, T, C, and G. The order of these letters forms the DNA sequence.

What are Genes?

Sections of DNA with instructions for proteins are called **genes**.

The *F8* gene makes the factor VIII protein.



What is a DNA Variant?

DNA differences are called **variants**. There are different kinds of DNA variants.

Single Nucleotide Variant

A change of one letter is called a **single nucleotide variant**.

Deletion

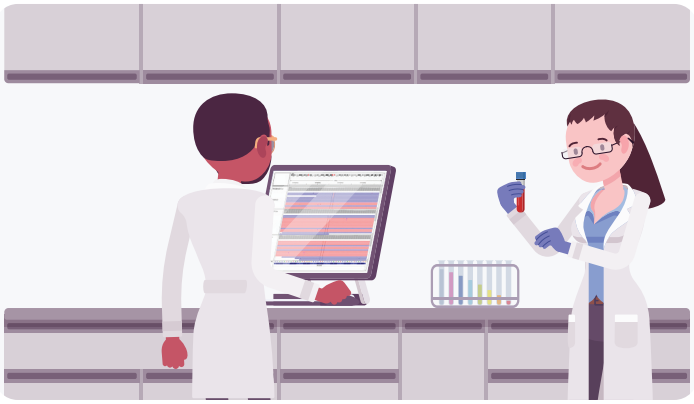
A missing sequence is called a **deletion**.

Insertion

An extra sequence is called an **insertion**.

Inversion

When a DNA segment breaks off and reattaches itself backwards, that's an **inversion**. *F8* inversions are the most common cause of severe hemophilia A.



What is a Genotype?

A **genotype** is a DNA variant that causes disease or changes something else about a person, like eye color.

Genetic Testing

A hemophilia genotype can be found in >98% of people with hemophilia A. Testing must be done in experienced laboratories. Special detection methods are required for some kinds of variants that cause hemophilia, such as *F8* inversions.

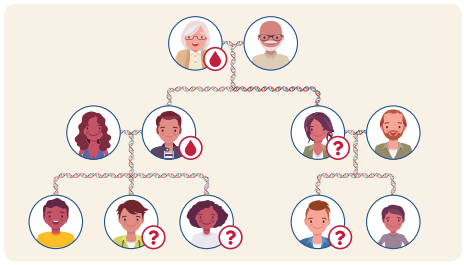
Genetic Counseling

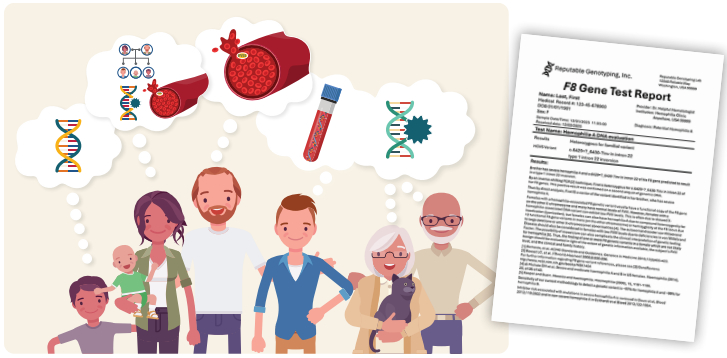
Genetic counseling is recommended for anyone who receives genetic testing.

Genetic counseling helps people

- understand how hemophilia might affect them,
- make sense of their test results, and
- think about what the results could mean for their family.

Genetic counseling also helps people prepare for other possibilities, like discovering something unexpected that wasn't the reason for testing.





Genetic Test Results

The results of genetic testing are provided in a report. The laboratory may report:

- A genotype that clearly causes hemophilia
- A DNA variant where there is uncertainty as to whether or not it causes hemophilia
- That no genotype was detected.

Testing results should be shared with health care providers, family members, and anyone else who may need to know.

Where Can I Get More Information?

To find out more about the HARP:

https://harpf8.org/

To learn more about hemophilia:

https://www.ncbi.nlm.nih.gov/books/NBK1404/

To find a Genetic Counselor:

https://findageneticcounselor.nsgc.org/

https://harpf8.org

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