

# How Do I Know If I'm A Carrier of Hemophilia?

Females who may be carriers of hemophilia can be divided into two groups based on their family histories: obligate carriers and possible carriers. Genetic testing is sometimes necessary to determine whether a female is a carrier.

## Obligate Carriers

- Girls and women known to be carriers based on their family history alone
- A female is an obligate carrier if one of the following is true:
  - Her father has hemophilia
  - She has two or more sons with hemophilia
  - She has a son with hemophilia and at least one other relative with hemophilia (such as a grandfather, brother, uncle, grandson, nephew, or cousin)
- Obligate Carriers do not require genetic carrier testing

## Possible Carriers

- Girls and women who have a chance of being carriers for hemophilia, but their carrier status cannot be determined by family history alone
- Possible carriers need genetic carrier testing to determine their carrier status

## How is genetic carrier testing performed?

- Individuals with hemophilia have a change within their factor VIII or factor IX gene that has caused it to make lower-than-normal amounts of clotting factor VIII or clotting factor IX
- In order to perform carrier testing, it is preferred that a male family member with hemophilia be tested first to identify the genetic alteration in the factor VIII or factor IX gene causing hemophilia in the family
- Once the familial genetic alteration has been identified, female family members who are possible carriers can have a blood test to look for the presence or absence of this specific alteration within her factor VIII or factor IX genes, determining her carrier status

## None of the males with hemophilia in my family have had genetic testing. Can I still be tested?

- Yes, but we would suggest you talk to your family members who have hemophilia to see if they would be willing to have genetic testing first.
- If a male family member with hemophilia is unavailable or unwilling to be tested, the second choice would be to perform genetic testing for an obligate carrier in your family first.
- If neither a male with hemophilia nor an obligate carrier in your family are available, a possible carrier can still have genetic testing performed. This, however, does not provide the most accurate results. There is a small chance of receiving a false negative result. This can be discussed further with a genetic counselor.

## How can I get more information?

For additional information, to discuss your specific family history (including your chance to be a carrier), obtain more details about genetic testing, or to pursue genetic testing, please contact one of the IHTC's genetic counselors.