

# Chapter 4

## GENETIC ASSESSMENT

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### Genetic Assessment: **WHAT YOU NEED TO KNOW**

#### Who should receive genetic testing of hemophilia?

People with suspected or diagnosed hemophilia

And their 'at risk' female family members

#### Why is genetic testing done ?



To define specific genetic mutation



To establish diagnosis in difficult cases



To predict risk of inhibitor development



To identify female carriers



To provide prenatal diagnosis

#### How does genetic testing work?

##### **1** Pre-test Genetic Counselling

A genetic counsellor will provide information on the testing procedure and possible consequences of the test results

##### **2** Genetic Testing

Genetic testing is done through a blood test

Coagulation factor levels and von Willebrand factor activity will be tested

##### **3** Test Results & Post-test Genetic Counselling

Everyone being tested should receive post-test counselling to learn about the results and determine a course of action