Chapter 4
GENETIC ASSESSMENT

Megan Sutherland, Carlos De Brasi, Barbara A. Konkle, Shrimati Shetty, Glenn F. Pierce, Alok Srivastava

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