

## Genomic testing – Patient information sheet

### Why have I been offered genomic testing?

Your healthcare professional thinks that there may be a genetic explanation for the symptoms you or your child have. Genomic testing can help diagnose a condition quicker and more accurately and may reduce the number of medical procedures a person requires. The results of testing can help tailor medical care, such as treatment and surveillance.

### What is genomic testing?

This test aims to identify changes (known as variants) in your genes that can affect your health.

Genomic testing is usually performed on a blood sample, but saliva or tissues samples may be tested. DNA (our genetic material) is extracted and the code is read to look for variants.

This test may look at a single gene, a small group of genes or large number of genes. Variants that are identified will be examined to determine whether they fit with the reason you were offered genomic testing.

### What could my genomic test results show?

There are four possible outcomes from your genomic test;

- **Gene variant identified.** This would confirm a diagnosis for you and would clarify the potential implications for you and your relatives. Referral to Clinical Genetics would be recommended to discuss these implications further and to support you in sharing information with family members.
- **No gene variant identified.** This result would not confirm the cause of your symptoms and does not exclude the possibility that there is an underlying genetic cause for your symptoms. There are limitations with genomic testing and the current technology. Further testing may become available in the future.
- **Variant of unknown significance identified.** These are variants in the gene that the laboratory is uncertain would cause the symptoms you have. This result would not confirm a diagnosis for you. Further investigations may be offered to you or you may be advised that this gene change could be reviewed in the future, as more information may become available.
- **Unexpected finding.** A variant identified in a gene that is not related to the reason you were offered testing. These are sometimes referred to as incidental findings.

### **How will I receive my results?**

Your result will be returned to your healthcare professional, who will contact you to explain these. You should discuss with them how you will be informed about your result. Once you receive your result you may be referred to Clinical Genetics or other specialist for further advice.

Genomic testing can take weeks to months to complete. You will be advised how long your test may take.

### **Why might genomic testing be important?**

For you

- Identifying the genetic cause of a condition can help patients understand how they have developed the symptoms they have.
- It may help provide more accurate information about how the condition may develop in the future.
- It may clarify treatment options.
- Access to support groups, with opportunities to connect with others with the same condition
- There may be family planning options that you want to consider.

For your relatives

- It may confirm whether other family members could get the condition.
- Relatives could seek advice about screening and treatment options that may be available to them.
- Family planning options

### **Insurance and genomic testing**

When applying for some types of insurance you are required to disclose medical information about you and your family. If genetic testing confirms a diagnosis for you, this result would have to be shared to an insurance company when obtaining or updating an insurance policy. Your results will not be disclosed to insurance companies without your consent.

Further information is available at [www.abi.org.uk](http://www.abi.org.uk)

### **Use of your data/result**

Your genomic test results may be shared with other healthcare professionals involved in your care, so that they may provide you with the appropriate on-going care.