Inherited Metabolic Disease Service

Genetic testing

What are genes?
Your body is made up of trillions of cells, each of which contains the genetic material that determines the characteristics that make you a unique individual. We all have numerous differences in our genetic code and whilst some changes are responsible for characteristics such as eye colour others can cause disease or sometimes don’t appear to have any significance at all.

Your genetic code is made up of molecules of DNA that are grouped together to form a gene. Each gene provides the instructions for your cells to make specific proteins. Hundreds and thousands of genes join together to create chromosomes. You have 23 pairs of chromosomes, having received one copy of each from your parents.

What is a genetic test?
A genetic test is a type of medical investigation that identifies changes in your chromosomes and/or genes.

Why do I need this test?
☐ This is to help understand what is causing your symptoms
☐ This is to establish whether you are at risk of developing a disease
☐ This is to help confirm a diagnosis in a family member

What do I need to consider before having this test?
There are some things you should take time to consider before having this test:

- Sometimes test results might be inconclusive.
- The results might affect your ability to get life insurance/ medical insurance.
- It can take some time before the results are available and this can be a source of anxiety for some people.
- Sometimes genetic testing can show that family members may not be genetically related
- How will you deal with the results?
- How will you tell other members of your family?
- Why do you want to know?
- What impact will the results have for you when considering whether to start a family.
What does the procedure involve?

A blood sample is collected from you and then sent to our molecular genetics laboratory. There are different types of tests the laboratory can then perform:

**Chromosomal studies**: this looks at whether you have the full complement of chromosomes and is also known as karyotyping. Slightly more detailed studies known as fluorescent in situ hybridisation (FISH) analysis can identify whether the chromosomes are complete or whether they have any missing or additional sections, and chromosomal microarray analysis can be used to identify very tiny changes in chromosomes.

**Single gene test**: this test is used if a specific disease is suspected. The responsible gene is identified and examined for any potentially disease causing changes in the DNA sequence.

**Disease specific panel**: this test is used if a particular set of symptoms have a variety of different genetic causes. All of the potentially responsible genes that are included in the panel are identified and examined for any potentially disease-causing change in the DNA sequence.

**Whole genome/exome sequencing**: these tests, which look at all of your genes, are used if it is unclear which gene might be causing the symptoms. Your genetic material is examined for any potentially disease causing change in the DNA sequence.

When will I get the results?

Genetic testing is complex and can take some time to complete. Identifying a DNA change is a little like looking for a spelling mistake in one word, in one line, on one page, in one chapter, of one book, in one section, on one floor of a large library! Results usually take between 4 to 16 weeks, although this can take much longer.

Understanding the results?

The test results may show:

- You have a change in your genes which is known to cause a health condition.
- You do not have a change in your genes which is known to cause a health condition.
- It’s not clear what the results mean for your health (but doctors may have a better understanding of the results in the future) - this is sometimes termed a variant of uncertain significance (VUS).

Please tell the doctor or nurse whether you would prefer to receive your results in a letter, by telephone or at a clinic visit.
What next
If you would like to proceed do confirm this with your healthcare team and state whether you will be happy to share your results to help other family members if necessary.

Please tell your doctor or nurse if you would like to take a little longer to decide about this.

Further Information
Further information can be obtained from the Genetic Alliance support group, or your local genetic counselling service.
www.nhs.uk/conditions/genetic-and-genomic-testing/

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Document history
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Contact number: 01223 274634
Publish/Review date: December 2019 / December 2022
File name: Genetic testing_V1.doc
Version number/Ref: 1 / Document ID 101389