NHS
London North
NHS Genomic Laboratory Hub

Information on Whole Genome Sequencing for patients

As part of your care, your doctor may offer you various tests to try and identify the genetic cause of your condition. Your doctor will discuss these with you in greater detail at your appointment. One of these tests is called Whole Genome Sequencing (WGS).

What is Whole Genome Sequencing (WGS)?

Your genome is your body's 'instruction manual' and contains nearly all the information needed to create, run and repair you. Your genome is made up of a chemical code, called DNA, which is like a series of 'letters' that can be looked at using a technique called sequencing. WGS reads through all 3.5 billion letters of your DNA and is what we call a 'genomic test'. WGS is one of the tests you may be offered to see if we can find a change in your DNA that might be the cause of your condition. In some cases we may need to compare your genome with your family members' DNA to help us identify harmless changes which can run in families as distinct from those which may be causing your condition.

What results might you get?

- **No relevant findings** - This means that we have not identified a cause for your condition. In the future, as knowledge and technology improves, we may be able to find the cause and we will discuss when you should seek further advice, for example if there is a change in the condition or if someone in the family is planning a pregnancy.
- **Relevant finding** – This means there is a change which clearly explains your condition. This may influence your treatment or inform your family members of their risk of having the same condition.
- **Uncertain finding** - This is a change which could explain your medical condition, but more tests or research may be needed to determine if this is relevant. This might include testing other family members. In some cases, this could be a change that we cannot be sure whether this is the cause of your condition or just part of normal variation. This might become clearer with time and as our knowledge of the genome improves.
- **Incidental finding** - The test may reveal an unexpected change in your genome which is not related to your condition but may have health implications for you or your family.

As with all health data, the data from your genomic test will be stored securely within the NHS so that we can reanalyse it in light of new knowledge and understanding.

What will happen if I am offered WGS?

Your doctor will explain the test in more detail, why you are being offered it and how you will get your results. You will be asked to sign a consent form if you agree to the test, understand what results you might get and when. If you agree to have the test, you will be asked to provide a blood sample. As this is a complex test, results are usually available after a few months.

Contributing to research

Whole genome sequencing is a new and powerful technology that is helping us understand the cause of many conditions, how they develop and what it means for people with those conditions. In many cases, once we understand the underlying genetic cause we can work with researchers in our universities and industry to develop new treatments.

To help us continue these developments, everyone that has WGS will be offered the opportunity to contribute their anonymised data to a national data store that can be accessed by academic or commercial researchers. Access will be granted and monitored by a committee made up of doctors, clinical academics and patient representatives and no identifiable data will be released. We will also ask your permission for us to contact you should any relevant research studies become available.

Your doctor will discuss this in more detail and ask you to sign a form to say whether you agree to donate your data to research or not. Contributing in this way may help you or your family in the future, and may help others as well.

If you decide not to have WGS or not to contribute your data for research, you will still receive the best possible care from your doctor. The same applies no matter what the result of your WGS may be, should you choose to have the test.

For more information about WGS

We have developed two short videos to explain genome sequencing in more detail. The first video explains what a genome is, how genes can cause health problems, and what genome sequencing is:
My Genome Sequence:
http://bit.ly/mygenomesequence

The second video looks in more detail at how genome sequencing is done and describes some of the current limitations and uncertainties with the technology:
My Genome Sequence Part 2:
http://bit.ly/mygenomesequence2

You may also want to visit the Genomics England website at
https://www.genomicsengland.co.uk/understanding-genomics/