**What happens when I go for whole genome sequencing?**

**Genomics 101 explainer episode transcript**

**Florence: What happens when I go for whole genome sequencing? I'm joined by Katrina Stone, Clinical Genetics Doctor, to find out more. So, Katrina, first things first. What is the purpose of whole genome sequencing?**

**Katrina:** The purpose of whole genome sequencing is to try to make a precise genetic diagnosis for someone with a suspected or confirmed genetic condition.

**Florence:** **And why might someone get whole genome sequencing?**

**Katrina:** They might get whole genome sequencing because they are known to have a condition which is likely to be genetic, but the medical team wants to find out what the exact genetic cause is. In other cases, the diagnosis might not be known, and the reason for doing whole genome sequencing is to find out whether there is a genetic condition present.

Some of the benefits of having the test is that. If a condition is identified, this can provide an explanation for the family about what's been going on, and it can also bring to an end further unnecessary investigations. Also, if a genetic diagnosis is confirmed, this can sometimes point towards other things which might need to be kept an eye on for the individual.

In addition, once a diagnosis is confirmed, a doctor can advise the family on the likelihood of other members of the family or future children being affected with the same condition, and they can use this information to help with future family planning.

**Florence:** **So, then what happens when a person physically goes to get the test?**

**Katrina:** In most cases, an individual will see a specialist doctor. This might be a genetics doctor, but it could be a doctor specialising in another body system. They'll do a full assessment of the individual, including finding out lots of information about them and their family, and also examining them to look for any clues that might point towards a specific genetic diagnosis.

Once the family have decided to go ahead with the test, their consent will be taken, where the test will be explained in more detail, including the pros and cons of going ahead with the test and after that samples can be taken. Usually this is a blood sample, but occasionally a saliva sample or cheek swab could be taken.

The best way to perform whole genome sequencing is with a sample from the person being tested along with both of their parents. And the reason for this is that it makes it easier to separate out genetic changes that are more likely to be significant from those that just represent harmless genetic variation what makes us all unique.

**Florence:** **What happens to this sample after the test has taken place?**

**Katrina:** So, the blood samples will go to a genetics lab where the genetic material known as DNA is extracted. The DNA is then sequenced, so we get an electronic file of all their genetic information. This is then analysed firstly by a computer which picks out changes or variants in their DNA, which are more likely to be significant.

After this, a trained clinical scientist analyses the data in detail. Sometimes there isn't a clear-cut result, and the scientists might need help from others and interpreting the result, but if there is, they can create a report which details the likely diagnosis.

**Florence:** **And finally, how will the patient get the result from their whole genome sequencing test?**

**Katrina:** Usually, the result is fed back to the patient and their family by the clinician who arranged their testing or one of their close colleagues. It's important to note that not everyone will get a genetic diagnosis from the test. This doesn't necessarily mean there isn't a genetic diagnosis present.

There are several reasons why tests might be negative. One is that no test is perfect and something important might have been missed because of the way the test works. Or it may be that the person being tested has a change in a gene that hasn't been described as causing a disease before, so we wouldn't even know to look for it.

There's also a possibility that there isn't a single genetic cause for their symptoms. Rather, lots of minor genetic factors are causing their condition. We're not very good at testing for these yet. Finally, there could be a non-genetic cause that just hasn't been identified yet.

One of the benefits of having a whole genome sequencing test is that the data can be stored and looked at again in the future, either in light of new evidence or once our knowledge of genetics has improved.

**Florence:** **That was Katrina Stone explaining what happens when you get whole genome sequencing. If you'd like to hear more explainer episodes like this, you can find them on our website at** [**www.genomicsengland.co.uk.**](https://www.genomicsengland.co.uk)

**Thank you for listening.**