Genomics 101 transcript: Genetics Vs genomics, What’s the Difference?

Dr Rich Scott

**Naimah:** Do you know the difference between genetics and genomics today? I am joined by Dr Rich Scott, who is the Chief Medical Officer and Deputy CEO here at Genomics England, to help explain the difference between these two closely related fields.

So, Rich, what is the difference between genetic and genomic testing?

**Rich:** I think the first thing to say is that people should not worry too much about exactly which word they use, and we will come back to why that is, but also what the differences are between the two. So, overall genetic and genomic testing are about testing your DNA. Just to remind people, DNA is the chemical code that's made up of four different chemical alphabet letters, A, T, G and C, that together make up your whole genome – 3 billion letters of that code that tell your body how to grow and develop and look after itself and make you, you. Genetic and genomic testing are ways in which you test your DNA to help understand about your health and how you can be looked after best.

**Naimah:** So, can we go a bit deeper into what is genetics and what is genomics?

**Rich:** Going back to your genome and what all of that DNA is, as I say, your genome is the 3 billion letters that make up your full genetic code and that's made up of different components. The bit that we understand best, and we focus on most, are the genes, and those are the sections of DNA that tell the body how to make RNA and then proteins, the building blocks of the body; for example, enzymes that do particular jobs that look after different processes in your cells, and so forth. So, those genes are particularly important. They actually only make up about 1 or 2% of the whole of your genome.

When we are talking about genetic testing, we are talking particularly about the testing that looks at those genes. When we are talking about genomic testing, what we are talking about is looking more broadly across the genome, whether that is, for example, to look at things particularly in the gaps between the genes or whether it's to understand what's going on more generally. Just going back to those words again, I think people should not worry if they accidentally interchange them or they can't quite think which sort of test it is. If you say genetic or genomic testing, you are talking about DNA testing, and it is broadly the same thing.

**Naimah:** When you're looking at a single gene compared to a larger piece of DNA, can you give me some examples of what this might tell us?

**Rich:** Yes, so when one's talking about genetic tests, as I say, you're mainly talking about testing, looking at particular genes, so the sort of situation where that's used and a nice example would be, for example, a family where who has breast or ovarian cancer, and there's a strong family history of breast and ovarian cancer, to help guide the care of their relatives. It is useful to look at particular genes that are linked in a small number of families to having a high risk of breast and ovarian cancer. In that setting, you are using testing to look at very particular parts of the genetic code and particular genes to say, can we see any differences there that would explain that? So, that's a nice example of a genetic test. Thinking about how genomics testing is used, a nice example is thinking about using whole genome sequencing, which now is available routinely in the NHS in England to look into certain rare condition settings and also in certain cancers to look across the genome, look across all of the genes and, often, at bits in between, to help understand how to look after that person best. So, for example, in cancers, you can see patterns of misprints that have arisen in the tumour itself that can help you understand how to best guide and look after that patient.

**Naimah:** How long does it take to look at individual genes in comparison to larger pieces of DNA?

**Rich:** Most of the difference is how complex the process of understanding what is going on in the sample is. There are differences in exactly how long it takes to run the test in the lab, but actually those differences aren't very big nowadays. So, the testing itself, it is often more about the sort of complexity of the analysis, but there's not necessarily a big difference between the two.

**Naimah:** Can both of these be used for precision health?

**Rich:** Precision health is a term that's often used to say that by using certain tests, including genetic and genomic DNA testing, you can be more precise about how you give someone guidance, or you decide what treatment pathway is best for them. Both genetic and genomic testing can help you with that. For example, helping to identify which rare condition someone has or to classify someone's tumour better can help you decide how broadly to advise them, or what sort of follow up they should have, and in some cases also identify quite precisely particular medicines that might be helpful.

**Naimah:** You mentioned before that genomic and genetic testing can be used in routine care, can you explain a bit more about this?

**Rich:** Yes, testing of DNA has been around for years now. In the UK, for example, NHS genetic testing labs have been around for quite some time. In the last few years there has been a real advance in terms of the amount of testing that is possible and available and the breadth of the settings in which it can be used. At the moment the major focus is on testing where people have particular clinical features that raise the possibility of a rare condition or certain cancers. Also, increasingly, and this is an area where things will be used more and more in the coming years, we think is Pharmacogenomics, which means testing the DNA to understand which medicine to pick for someone, whether that's because they would benefit from a different dose of that medicine or because there's a higher chance of them having a reaction to the medicine, and so helping to make those choices.

**Naimah:** That was Dr Rich Scott explaining the difference between genetics and genomics. If you have any questions, please feel free to email us at info@genomicsengland.co.uk. Thank you for listening.