Genomics 101 transcript: What is a genome?

**Naimah:** Here at Genomics England, you might hear us use a lot of complicated scientific terms, but today we're going to go right back to basics to help you understand a bit better about what we do here. Today I'm going to be speaking to Greg Elgar, who is the director of Sequencing in Research and Development here at Genomics England.

So first of all, Greg, can you tell me what exactly is a genome?

**Greg:** Your genome is all of your chromosomes in a cell stretched end to end, and then all the DNA along those chromosomes. So, it's a whole string of Gs, As, Ts and Cs, which are the four bases that make up our DNA sequence. So DNA is a very important part of our bodies, and almost every living cell in our body has two copies of our genome.

There are exceptions to that. So red blood cells, for example, don't have any DNA in them at all. But apart from that, pretty much every cell has a copy of our DNA, and that means each cell can work autonomously to make proteins from that copy of the genome.

**Naimah:** Okay. And what's the difference between a gene and a genome?

**Greg:** A gene is a unit of our genome, if you like. It's a small fragment that together can make proteins in our cell.

**Naimah:** What do we mean by a genome sequence?

**Greg:** So our genomes, as I've mentioned, are made up of four different bases, A, C, T and G. And it's that the order of those bases, so they can occur at any order. So you could have three A's in a row followed by a T and then a G, for example. Or you could have three G's followed by an A and a T. So genomes are made up by enormously long strings of those bases. And the order of those bases define what they mean in terms of how they get translated into our cells and into the way we look and our diseases and everything else. So, we like to define the sequence of a person's genome because once we've done that, we can see the differences between our genome and someone else's genome.

**Naimah:** Does everyone have the same genome, or how does that look?

**Greg:** So largely we have the same genome, but there are a number of differences and there are so many bases in our genome. So, there are 3000 billion bases that make up our genome. And so even if only a tiny percentage of those say 0.1% are different, that ends up being quite a lot of bases that are different.

Some of those bases are in parts of our genome. That means they have no consequence to us if they're different, because our genome is cluttered with lots of really cool genes, but also lots of bits that aren't actually that important or we don't really understand the true significance of them. So if they're different between you and me, that actually doesn't have any consequence.

But then there are other differences in our genome, and they are quite often those that fall within genes that do make a difference. Again, some might not, but some do, and some of those have really quite critical and important differences.

**Naimah:** So what do these differences look like?

**Greg:** So everything you can possibly imagine in the difference between one person and another, then the likelihood is that those differences are attributed to differences in our genome.

So a really catastrophic difference, for example, might be that one person gets cancer and one person doesn't, or one person gets a rare disease or has a rare disease. Alternatively, another difference may just mean that someone is slightly taller than someone else or has different colour hair or different colour eyes.

These are all fundamentally the same kinds of differences in the genome. It's just that depending on where they're placed means they have very different consequences.

**Naimah:** And do some of these differences affect how we might respond to medicine?

**Greg:** Yeah, absolutely. So one of the really big things at the moment is understanding that everybody's genome actually responds differently to different medicines. And that's because you may have a change in a gene that metabolizes or, or breaks down a particular drug or chemical. And that difference may mean that you break it down more slowly or more quickly, and that can mean that a drug can be more or less effective, for example.

**Naimah:** Having said that, does everything that we see about a person is that based on their genome?

**Greg:** Well, a lot of what we see about a person is based on their genome, but we have a lot of influence over the way we look and the way we act. And, and a good example of that is that there may be some genes or some sequence that defines how you respond to eating. So, some people have a greater tendency to become overweight, even if they eat the same amount as someone else, and that's defined by their genes.

But we can also control how we look by eating less, for example, or exercising more. So, although the genome provides a blueprint of how we function and grow and develop and look, we can also influence that by our lifestyle, medicines and various other things that will interact either directly with our genome or indirectly because we overcome some of the variations in our genome.

**Naimah:** And can you give me an example of one of these external factors that you're talking about?

**Greg:** Yeah, so I suppose the easiest one to describe is the way we interact with the environment and how that might lead us to get cancer. So a good example is skin cancer. We may all have different predisposition to skin cancer based on our genome. So some of us might be more likely to get skin cancer because of our gene sequence, and some people may be less likely. However, if we don't expose ourselves to too much sunlight and too much UV light, then even if we have a strong predisposition to skin cancer, we will probably never get it because we're not exposing ourselves to environmental factors that can interact directly In this case with our genome and our DNA sequence.

**Naimah:** That was Greg Elgar explaining what is a genome. If you've got any questions, please feel free to email us on info@genomicsengland.co.uk. Thank you for listening.