National Genetics and Genomics Education Centre

Variations in DNA and their effects

We're all different. We look different, we act differently, we think differently. Everybody is an individual. Even with identical twins, once you know them well enough, you realise they're different. But we're all human, and despite those differences, when medical students learn about anatomy and physiology, what they learn applies to pretty much all of us. Underneath all those individual differences, there's a basic human anatomy and physiology that, barring rare accidents and diseases, we all share.

It's the same with our DNA. Each of us has our own DNA, which is individual and has differences from everybody else's. That's why DNA is so powerful in criminal investigations, or in checking paternity. But underneath all those individual differences, there's a general similarity, as confirmed by the Reference Human Genome Sequence that was the result of the Human Genome Project.

For biologists wanting to know how humans work, what matters is those common features that define us as human. But geneticists are especially interested in the individual differences, particularly in DNA.

DNA variants can be grouped into four categories:

- Most of the many individual differences have no effect whatsoever. They don't affect your health, your appearance, your personality or anything else.
- Some variants are responsible for the individual differences between normal healthy people – differences in hair and eye colour, blood group and so on.
- Some variants affect your natural susceptibility to common diseases such as diabetes, heart attacks or schizophrenia. Whether or not you actually get any of these diseases depends on a lot more than your genes – your lifestyle, your environment and simple bad luck play a large part. But particular DNA variants make some people more or less likely to develop the disease, given a particular environment and lifestyle.
- Some variants directly cause a disease. These are the genetic diseases like cystic fibrosis or muscular dystrophy. For these rare diseases, the cause is a failure of one or more copies of a particular gene (different for each disease) to work correctly, as a result of an alteration in the DNA code of that gene.

Most of your DNA is determined at the moment of conception. You inherit a particular set of variants from your father and another set from your mother, and those are the variants that make your DNA individual and different from anybody else's. These are your constitutional variants. Your brothers and sisters will have inherited a different selection of the variants your parents have, so everybody is individual. Sometimes however a new variant arises in one of your cells as a result of some accident affecting just that cell's DNA. These are called somatic variants. Constitutional variants are inherited, and passed on from parents to children, but somatic variants are restricted to the body of that particular individual. Mostly they are unimportant, but geneticists are interested in them because sometimes they can set a cell on the road to becoming a tumour.