We all have hundreds of DNA flaws, UK geneticists say

By Helen Briggs
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Everyone has on average 400 flaws in their DNA, a UK study suggests.

Nobody is perfect from a genetic perspective

Most are "silent" mutations and do not affect health, although they can cause problems when passed to future generations.

Others are linked to conditions such as cancer or heart disease, which appear in later life, say geneticists.

The evidence comes from the 1,000 Genomes project, which is mapping normal human genetic differences, from tiny changes in DNA to major mutations.

In the study, 1,000 seemingly healthy people from Europe, the Americas and East Asia had their entire genetic sequences decoded, to look at what makes people different from each other, and to help in the search for genetic links to diseases.

The new research, published in **The American Journal of Human Genetics**, compared the genomes of 179 participants, who were healthy at the time their DNA was sampled, with a database of human mutations developed at Cardiff University.

It revealed that a normal healthy person has on average about 400 potentially damaging DNA variations, and two DNA changes known to be associated with disease.

> **All of our genomes contain flaws; some of us will carry deleterious variants but will not be at risk of acquiring the associated disease for one reason or another.**
> Dr Chris Tyler-Smith, Wellcome Trust Sanger Institute

"Ordinary people carry disease-causing mutations without them having any obvious effect," said Dr Chris Tyler-Smith, a lead researcher on the study from the Wellcome Trust Sanger Institute, Cambridge.

He added: "In a population there will be variants that have consequences for their own health."

The research gives an insight into the "flaws that make us all different, sometimes with different expertise and different abilities, but also different predispositions in diseases," said Prof David Cooper of Cardiff University, the other lead researcher of the study.

"Not all human genomes have perfect sequences," he added. "The human genome is packed with pervasive, architectural flaws."

Personalised medicine

It has been known for decades that all people carry some genetic mutations that appear to cause little or no harm.

Many are only damaging if they are passed on to children who inherit another copy of the faulty gene from the other parent.

In others - around one in ten of those studied - the mutation causes only a mild condition, appears to be inactive, or does not manifest itself until later life.

Databases of human mutations, like the one at Cardiff University, will have increasing importance in the future, as we move into the era of personalised medicine.

More people have access to genetic information about themselves, with various companies offering screening of selected gene changes via the internet.

Meanwhile, the cost of sequencing a whole genome is dropping rapidly.

In the case of the 1,000 genomes study, samples were anonymous, and participants will not be given information about any gene changes linked with disease.

But as DNA sequencing becomes more widespread, ethical dilemmas will arise about what to tell people about their genes, especially when many risks are uncertain.

Dr Chris Tyler-Smith said: "All of our genomes contain flaws; some of us will carry deleterious variants but will not be at risk of acquiring the associated disease for one reason or another.

"For others, there will be health consequences, and early warning could be useful, but might still come as an unwelcome surprise to the participant."

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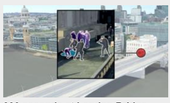
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