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By Sam Wong

**Study finds five new genetic variants linked to heart disease**

**Five new genetic variants linked to heart disease have been identified in a meta-analysis of four large genome-wide association studies, published in** [**Nature Genetics**](http://www.nature.com/ng/journal/vaop/ncurrent/full/ng.782.html) **this week.**

**The findings will guide research into new treatments for coronary heart disease, which kills 88,000 people in Britain each year.**

The discoveries add to 11 common variants previously shown to be associated with heart disease, and provide further evidence that many genes have a small but significant effect on heart disease risk.

The Coronary Artery Disease (C4D) Genetics Consortium, co-led by groups at Imperial College London and the Universities of [Oxford](http://www.ox.ac.uk) and [Cambridge](http://www.cam.ac.uk), compared the DNA sequences of thousands of people with heart disease in European and South Asian populations with controls from the same ethnic groups.

All of the variants linked to heart disease in the study appeared to be equally significant in people from European and South Asian ancestry.

Although several genes have been found to affect heart disease risk previously, the heritability of the disease suggests that more genetic factors are yet to be discovered.

The researchers noted that the magnitude of the effect of each new gene discovered is getting smaller, suggesting that there are not common variants with large effects waiting to be discovered, but rather a large number of variants with small effects contribute to heart disease risk.

[Professor Jaspal Kooner](http://www1.imperial.ac.uk/medicine/people/j.kooner/), [Dr John Chambers](http://www1.imperial.ac.uk/medicine/people/j.chambers/) and [Professor Paul Elliott](http://www1.imperial.ac.uk/medicine/people/p.elliott/) co-led the research at Imperial, and determined the contribution of these variants to heart disease amongst South Asians living in the UK.

“These findings add weight to the idea that a large number of genes affect your likelihood of developing heart disease, each gene having a relatively small effect,” Professor Kooner said.

“This means that genetic tests are unlikely to be useful for predicting heart disease, but each gene we discover tells us about the biological mechanisms underlying heart disease and gives us a new lead to look for new treatment strategies.”

The initial discovery phase comparing around 15,000 people with heart disease with a similar number of controls found 50 locations on the genome where single-letter variations in the genetic code were associated with disease.

These variations act as pointers to regions of the genome where genes involved with disease are likely to be found.

After replication studies in separate groups of participants to check the statistical significance of these associations, five new regions were found to be linked to heart disease.

The researchers examined tissue samples to see which genes in the regions they identified are more active in people with heart disease.

In two of the regions, they were able to find specific genes that were implicated in heart disease.

A genetic variant on chromosome 11 was linked to a gene called PDGFD, which the researchers found was overactive in tissue samples from arteries of the group who developed heart disease before reaching 60.

PDGFD encodes a protein that is thought to stimulate atherosclerosis, the development of fatty plaques in arteries which causes heart attacks and strokes.

Another variant on chromosome 10 was linked to a gene called LIPA, which was found to be overactive in samples of liver tissue from heart disease patients.

This gene regulates white blood cells called monocytes which play an important role in the formation of plaques.

The mechanisms linking the other three genetic regions with heart disease risk are still unknown, but their locations give scientists a starting point for future research.

The research was supported by funding from the [British Heart Foundation](http://www.bhf.org.uk), the [Wellcome Trust](http://www.wellcome.ac.uk), the [Medical Research Council](http://www.mrc.ac.uk), the [National Institute for Health Research](http://www.mrc.ac.uk), and the European Union.

Professor Kooner and [Dr John Chambers](http://www1.imperial.ac.uk/medicine/people/j.chambers/) from the [School of Public Health](http://www1.imperial.ac.uk/publichealth/) at Imperial lead the [LOLIPOP](http://www.lolipopstudy.org/introduction) study, involving over 30,000 volunteers from West London, which contributed data to the analysis.

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