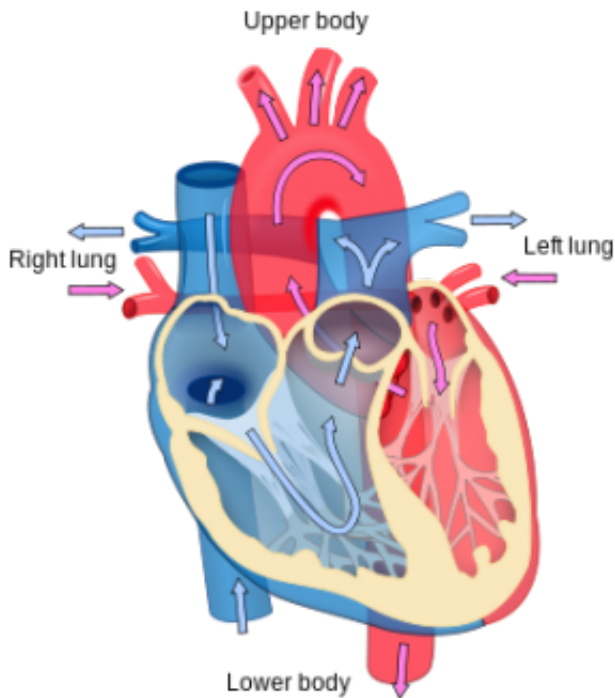


Researchers answer important scientific debate connected to heart disease

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Heart diagram. Credit: Wikipedia

Researchers from the University of Ottawa Heart Institute (UOHI), together with the teams of Dr. Martin Farrell at Oxford University, and Dr. Sekar Kathiresan at the Broad Institute, have found the answer to an ongoing debate in the cardiovascular scientific world. Dr. Ruth McPherson and Dr. Majid Nikpay, researchers at the UOHI's Ruddy Canadian Cardiovascular Genetics Centre, report that the genetic basis of heart disease is largely derived from the cumulative effect of multiple common genetic variants, rather than from a few rare variants with large effects.

The study, published today in the eminent *Nature Genetics*, used the data from the 1000 Genomes project in order to obtain information on close to 10 million genetic variants (called SNPs). The analysis involved 60,000 [heart disease](#) patients, 120,000

healthy individuals, from a total of 48 studies around the world. Not only is the number of genetic variants much greater than the approximately 1 million previously studied, this is the first time that researchers have been able to study the link of rare genetic variants present in as few as 1 in 1000 people at risk of heart disease.

"Our analysis provides a comprehensive survey of the fine genetic architecture of coronary artery disease (CAD), showing that genetic susceptibility to this common disease is largely determined by common SNPs of small effect size rather than just a few rare variants with large effects," say the authors of this important study.

Dr. Majid Nikpay, post-doctoral fellow at the Ottawa Heart Institute, also used an alternative statistical method of analysis to find two new risk markers that have an effect only if an individual has inherited two copies of the "bad gene", that is one from each parent. In addition to discovering a total of 10 new risk markers, by using other statistical approaches, this research team has produced a list of 202 genetic variants in 129 gene regions that together explain approximately 23% of the heritability of coronary heart disease as compared to only 11% reported in previous studies.

"Many of these genetic variants are likely to exert their effects on the walls of arteries, making them more susceptible to the common heart disease risk factors such as cigarette smoking, diabetes and cholesterol," added Dr Ruth McPherson, Director of the Ruddy Canadian Cardiovascular Genetics Centre at the University of Ottawa Heart Institute.

A number of preventative strategies target the [vessel wall](#) (control of blood pressure and smoking cessation), but the large majority of existing drug treatments for lowering CAD risk operate through manipulation of circulating lipid levels and few directly target vessel wall processes. Detailed investigation of new aspects of vessel wall biology

that are implicated by genetic association but have not previously been explored in atherosclerosis may provide new insights into the complex etiology of disease and, hence, identify new targets.

Provided by University of Ottawa

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