

How natural genetic differences can affect heart health

September 4 2019

The biggest risks for cardiovascular disease are smoking and poor diet. However, different people are more susceptible to heart disease based on very slight differences in their genes, called variants. While there have been many studies that have linked variants to cardiovascular traits, it's unclear whether these variants have functional consequences, like altered gene or protein expression. In a new study from the Cardeza Foundation for Hematologic Research at Thomas Jefferson University, researchers have discovered two slight gene variations that may modulate the behavior of platelet cells, and subsequently affect the risk of developing cardiovascular disease.

Normally, when platelets become activated, they stick together and clot wounds to stop bleeding. In disease, platelets can aggregate in response to triggers like unhealthy fats, and contribute to plaque formation in diseases like atherosclerosis. "Platelet numbers and volume are regulated by the expression of certain genes," says Leonard Edelstein, Ph.D., research assistant professor and senior author of the study. "If there's more of that gene, there are more platelets and a tendency to clot more." CD36 is one such gene that regulates platelet levels and activation.

In the study published in *PLOS Genetics* on July 25 2019, the researchers, including co-lead authors Namrata Madan, a postdoctoral fellow, and Andrew Ghazi, [graduate student](#), searched for small changes in the [genetic code](#) for CD36. The review of data identified 81 changes in the genome, two of which were functional variants, meaning they affected CD36 expression. When they used genetic editing to delete these

variants in cells, there was overexpression of CD36. This indicates that the genetic changes the researchers identified could be potential targets in modulating expression of CD36 and by extension platelet function. The researchers are now trying to identify what protein the variants they identified bind to, and the mechanism behind which it can regulate CD36 expression.

The study provides a paradigm for further testing of functional genetic variants and their relation to cardiovascular health. "We are now conducting an even more large-scale study testing 3,500 genetic variants, of which 150 are functional, and we want to investigate how these variants affect platelet function," says Dr. Edelstein. The results could potentially inform screening for genetic risk of [cardiovascular disease](#) and help develop targeted therapies.

More information: Namrata Madan et al, Functionalization of CD36 cardiovascular disease and expression associated variants by interdisciplinary high throughput analysis, *PLOS Genetics* (2019). [DOI: 10.1371/journal.pgen.1008287](https://doi.org/10.1371/journal.pgen.1008287)

Provided by Thomas Jefferson University

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