Medical Xpress

Common mutation linked to heart disease

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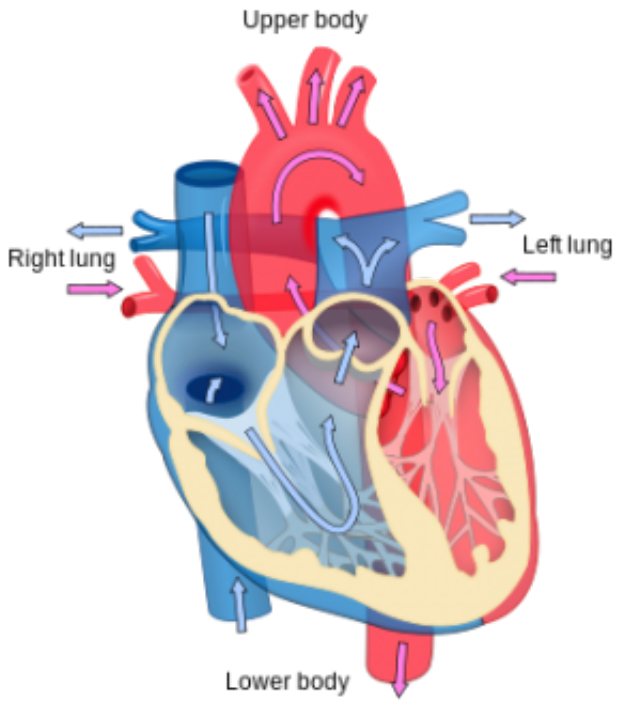

Heart diagram. Credit: Wikipedia

A common mutation in a gene that regulates cholesterol levels may raise the risk of heart disease in carriers, according to a new UConn Health study.

Researchers examined a mutation called the missense rs4238001 variant, which alters the type of protein made by the gene SCARB1, and thereby the body's cholesterol regulation.

The study, led by Annabelle Rodriguez-Oquendo, an endocrinologist at UConn Health, was based on information about more than 5,000 people who participated in the Multi-Ethnic Study of Artherosclerosis in major American cities from 2000 to 2002.

Rodriguez-Oquendo and her team charted the genotypes of the participants and tracked episodes of heart disease over a period of seven years.

They found that the mutation was associated with an increased risk of heart disease, particularly among men and African Americans, findings published in the 20 May issue of *PLOS ONE*.

The risk of heart disease among participants with the rs4238001 variant was up to 49 percent greater than the general population. Overall, men with the variant had a 29 percent higher risk than men without. African American males fared the worst, with the 49 percent increase risk, as compared to white males, whose risk was 24 percent higher.

The mutation is not rare, according to Rodriguez-Oquendo. A genetic test for the rs4238001 SNP is already available to help clinicians identify patients who are carriers so that they can offer counseling about heart risk prevention.

But now that the UConn study made the connection between the mutation in SCARB1 and heart disease, the researchers want to figure out a way to fix it.

"We want to go deep in the cell, and figure out how to repair it," Rodriguez-Oquendo says. "We're really interested in understanding more about how this protein gets chewed up and degraded faster."

The answers may impact the current standard of care for heart disease

prevention and treatment for patients who are carriers of the variant gene. That could happen through indirect means, such as adjusting hormone levels to alter cholesterol metabolism.

Provided by University of Connecticut