

Mutation protects against heart disease

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Human heart. Credit: copyright American Heart Association

According to new international research, just less than one per cent of the population is naturally protected against developing chronic coronary artery diseases. .

The *New England Journal of Medicine*, one of the world's most highly renowned health journals, has just published the results of international

genetic research collaborations.

Researchers from Iceland's deCode Genetics headed the project, which involved 292,000 participants of European origin, of which approx. 10,000 were from Denmark. Applying advanced gene sequencing techniques, the researchers located an area - a deletion - in the human genome, which lacked twelve DNA building blocks in 0.8 per cent of the participants.

Subsequent cell experiments revealed that due to the deletion, the serried gene - ASGR1 - is unable to establish the normal structure and function of the protein called the asialoglycoprotein receptor. The receptor protein binds certain sugars and surprisingly, it now turns out that the receptor plays an important role in our cholesterol metabolism and potentially related to vascular inflammation, and in whether or not we develop arteriosclerosis in coronary arteries.

"What's spectacular about the discovery is the fact that individuals with this rare and particular mutation have a lower level of cholesterol in their blood and their risk of developing arteriosclerosis is 34 per cent less. In other words, just under one per cent of the European population is fortunate to have been born with a mutation that decreases their cholesterol levels and thus to a certain extent protects them from developing coronary atherosclerosis," says Professor Oluf Pedersen, Novo Nordisk Foundation Center for Basic Metabolic Research, University of Copenhagen.

Potential for new preventive measures and treatments

"The mutated protein is expressed in a part of human biology, which we have not previously been focused on in our attempts to understand the mechanisms behind arteriosclerosis. This unexpected finding will undoubtedly result in many researchers examining the underlying

biological systems very thoroughly; hoping to utilize this new knowledge to develop new preventive measures and treatments for cardiovascular diseases," Oluf Pedersen elaborates.

Researchers from the universities of Copenhagen, Aarhus and Aalborg as well as the Center for Prevention and Health in Glostrup, Gentofte Hospital, Roskilde Hospital and Hvidovre Hospital represent Denmark in the study.

Provided by University of Copenhagen

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