

One-third of Greenlanders are at genetic risk for high cholesterol and cardiovascular disease

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A genetic variant that is present in nearly 30% of Greenlanders is linked to high cholesterol and an increased risk of cardiovascular disease,

according to a study published on June 9th in the journal *Human Genetics and Genomics Advances*. As noted by the authors, this is the first report of an association between this Arctic-specific variant, known as p.G137S, and cardiovascular disease.

"Due to its high frequency and large effect sizes, p.G137S has a marked population-level impact, increasing the risk of high cholesterol and cardiovascular disease for up to 30% of the Greenlandic population," says co-first study author Emil Jorsboe of the University of Copenhagen. "The variant is therefore a potential marker for early intervention in Arctic populations."

Cardiovascular disease is the number-one cause of death in many populations worldwide. It is tightly linked to elevated levels of low-density lipoprotein (LDL) cholesterol. The uptake of LDL-cholesterol particles from the blood into tissues such as the liver is mediated by the LDL receptor (LDLR). Mutations in the *LDLR* gene, which encodes this receptor, is a common cause of [high cholesterol](#) levels in the bloodstream.

Among Greenlanders, the prevalence of cardiovascular disease is likely to increase in the future due to increasing life expectancy and changing lifestyle. The common Arctic-specific *LDLR* variant, known as p.G137S, was recently shown to be associated with elevated [cholesterol levels](#). But until now, it was not clear whether this variant is also associated with an increased risk of cardiovascular disease.

In the new study, Jorsboe and his collaborators examined this possibility in a group of 5,063 Greenlanders. Roughly 30% of the individuals carried at least one copy of the p.G137S risk allele. Approximately 25% of the heterozygous and 55% of the homozygous carriers had high blood levels of LDL cholesterol.

Moreover, p.G137S was associated with an increased risk of ischemic heart disease, [peripheral artery disease](#), and coronary operations. Yet only a low proportion of individuals with very high levels of LDL cholesterol were receiving cholesterol-lowering therapy. In addition, elevated levels of LDL cholesterol for p.G137S carriers were independent of age, indicating that these individuals would benefit from [early intervention](#) and treatment.

"Our results showed that the p.G137S variant had an even larger impact on the lipid profile of Greenlanders than previously reported," Jorsboe says. "This variant further highlights the importance of carrying out [genetic studies](#) in smaller populations like the Greenlandic."

According to the authors, the results suggest that a screening program for the p.G137S variant could be highly useful for the early identification of individuals at increased risk for [cardiovascular disease](#), potentially improving preventive care and [public health](#).

"We think an intervention study would be of great importance, where one could investigate how cholesterol-lowering medicine improves the lipid profile in carriers of this genetic variant," Jorsboe says. "Defining the optimal treatment and prevention strategies for carriers of this variant is crucial, especially because one-third of the Greenlandic population are at risk."

More information: Torben Hansen, An LDLR missense variant poses high risk of familial hypercholesterolemia in 30% of Greenlanders and offers potential for early cardiovascular disease intervention, *Human Genetics and Genomics Advances* (2022). DOI: [10.1016/j.xhgg.2022.100118](https://doi.org/10.1016/j.xhgg.2022.100118). [www.cell.com/hgg-advances/full ... 2666-2477\(22\)00034-3](https://www.cell.com/hgg-advances/full...2666-2477(22)00034-3)

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