

A study of genetics of pericarditis increases understanding of newly approved drug treatment

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Rosa B. Thorolfsdottir, scientist at deCODE genetics in Iceland and first author on the paper. Credit: deCODE genetics

Sequence variants that protect against pericarditis have been discovered

at a genomic locus encoding interleukin-1 immune cytokines. A newly approved drug treatment for pericarditis inhibits these cytokines, according to a new study.

The article, titled "Variants at the interleukin-1 gene locus and pericarditis," was published today (Dec. 27) in *JAMA Cardiology* by scientists at deCODE genetics, a subsidiary of Amgen, and their collaborators from Denmark, U.S., and Iceland.

The study involves a genome-wide search for variants affecting the risk of pericarditis, a disease characterized by often painful inflammation of the fibrous sack surrounding the heart. A subset of patients experiences recurrent pericarditis that does not respond well to traditional treatment with unspecific anti-inflammatory drugs.

The role of specific immune processes in pericarditis is poorly understood and the aim of the study was to use [human genetics](#) to shed light on the pathogenesis of the disease.

The scientists found common variants in the genome that protect against pericarditis. They are located in a region with genes encoding interleukin-1 [inflammatory cytokines](#). Drugs inhibiting these cytokines have previously been used to treat other [inflammatory diseases](#), and they have recently been tested in clinical studies of recurrent pericarditis with good results. One of these drugs was approved by the US Food and Drug Administration for use in recurrent pericarditis as recently as 2021.

The results of the genetic study provide important insights. They suggest that interleukin-1 may be an important contributor to pericarditis in general, as the identified variants are common (up to approximately 50% frequency). Furthermore, the results provide the foundation for future studies, such as those aimed at understanding which interleukin-1 cytokines are most important and whether response to treatment is

affected by genotype.

More information: Variants at the interleukin-1 gene locus and pericarditis, *JAMA Cardiology* (2023). [DOI: 10.1001/jamacardio.2023.4820](https://doi.org/10.1001/jamacardio.2023.4820)

Provided by deCODE genetics

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