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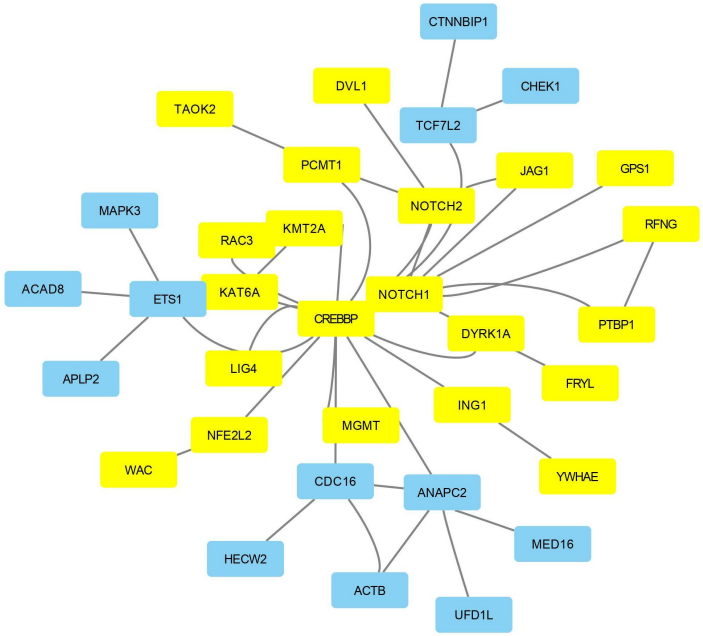
Kiel Researchers Discover Further CHD Genes

Research result supports individualized medicine

PRESS RELEASE

Berlin, September 14, 2021 - Researchers from Kiel have discovered additional genes involved in the development of congenital heart defects (AHF). The team led by human geneticist Professor Marc-Phillip Hitz of Schleswig-Holstein University Hospital in Kiel (UKSH) was able to make use of numerous previously published studies in its analysis thanks to a specially developed technique.



Alterations in NOTCH1 signaling pathway are involved in cardiac malformation. Figure created with Cytoscape version 3.8.2 (Shannon et al., 2003). © Marc-Phillip Hitz, UKSH

Kiel researchers report another milestone in research into the genetic causes of congenital heart defects. In a large meta-analysis, they discovered new genes involved in the development of congenital heart defects (CHDs). Thanks to a new method developed by UKSH bioinformatician Enrique Audain, the team led by human geneticist Professor Marc-Phillip Hitz was able to incorporate human genetic results from previously published studies into its comprehensive biometric study.

Seven Additional Genes Identified

In addition to the smallest losses in the chromosomes, so-called microdeletions, the researchers also discovered gene changes, new mutations, that occur for the first time

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and are involved in the development of a heart malformation. They analyzed data and samples from a total of 10,447 affected patients and 14,082 control subjects, including data and samples from the biobank of the National Registry of the Competence Network for Congenital Heart Defects. In whole genome analysis, 21 genes showed corresponding enrichment in the AHF patients. Fourteen of these had already attracted attention in previous studies due to their involvement in the development of congenital heart defects. For seven other genes - FEZ1, MYO16, ARID1B, NALCN, WAC, KDM5B and WHSC1 – this connection has now been demonstrated for the first time.

Impaired Communication Between Cells

The researchers also used the gene analyses to find out which protein networks and which interactions between them in the cells are most frequently associated with cardiac malformation: "In addition to disturbances in the Notch signaling pathway, through which the cells can respond to external signals during organ development, these are impairments in the DNA's own repair mechanisms and in the function of the cilia, those antenna-like projections on the cells that, among other things, control communication between the cells," explains human geneticist Anne-Karin Kahlert.

Discoveries for Patient-Oriented Precision Medicine

From the scientists' point of view, the results speak for the high importance of genetics in the development of congenital heart defects and support their future individualized treatment. "They enable better risk assessment regarding the recurrence of a heart defect within a family. And they serve us in developing more differentiated and effective treatment strategies for patients," summarizes Professor Marc-Phillip Hitz.

For Your Research

The results of the study conducted by researchers at Kiel University in cooperation with the Competence Network for Congenital Heart Defects, among others, appeared under the title "Integrative analysis of genomic variants reveals new associations of candidate haploinsufficient genes with congenital heart disease" in PLOS GENETICS in July.
https://pubmed.ncbi.nlm.nih.gov/34324492/

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