Researchers Identify New Gene Linked to Congenital Heart Defects

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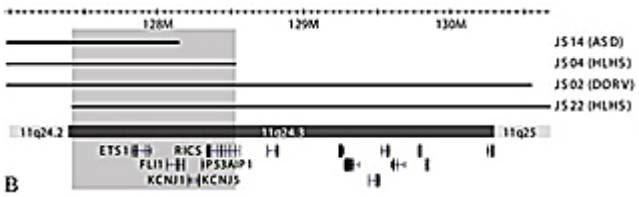
Researchers from the UC San Diego, School of Medicine and colleagues have identified a new gene, ETS-1, that is linked to human congenital heart defects. The landmark study, recently published online in the journal of Human Molecular Genetics, provides important insights into some of the most prevalent forms of congenital heart defects in humans, including ventricular septal defects and potentially hypoplastic left heart syndrome, a uniformly fatal heart abnormality.

Two siblings born with Jacobsen Syndrome treated by Paul Grossfeld, MD, of the UC San Diego School of Medicine.

"Identification of this gene may have implications for prevention of some of these most common types of congenital heart defects," said Paul Grossfeld, MD, associate professor of pediatrics for the UC San Diego School of Medicine and pediatric cardiologist for Rady Children's Hospital-San Diego.

The researchers performed high-resolution chromosomal micro-array mapping on human patients identified with the 11q- phenotype. Through a combination of human genetics and functional studies in genetically engineered mice, the researchers were able to pinpoint ETS-1 as a gene for causing at least a subset of the heart defects that occur in 11q- and the general human population.



Genetic mapping helped Grossfeld and fellow researchers pinpoint the region containing the ETS-1 gene,which is linked to congenital heart defects.

Characterized by a distinctive facial appearance, Jacobsen syndrome, also known as 11q terminal deletion disorder (11q-), is a rare chromosomal disorder resulting from the partial loss of one copy of human chromosome number 11. It is the loss of genes that leads to multiple clinical challenges associated with 11q- such as congenital heart disease, developmental and behavioral problems, low platelet counts, gastrointestinal, urinary and ophthalmologic abnormalities, failure to thrive and slow growth.

"Nearly 15 years ago, in my first month of pediatric cardiology training at UC San Diego, I came across my first patient with 11q-. To this day, I still care for this child," said Grossfeld. "Since then, I have devoted my career to learning everything about this syndrome and hope that we have now more clearly defined the path to understanding, and perhaps preventing, some forms of congenital heart disease."

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Researchers involved in this study include Maoqing Ye, UC San Diego; Chris Coldren, University of Colorado Health Sciences Center; Xingqun Liang, UC San Diego, Teresa Mattina, University of Catania; Elizabeth Goldmuntz, Children's Hospital of Philadelphia; D Woodrow Benson, Cincinnati Children's Hospital; Dunbar Ivy, University of Colorado Health Sciences Center; MB Perryman, University of South Dakota; Lee Ann Garrett-Sinha, SUNY-Buffalo; and Paul Grossfeld, UC San Diego.

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