Rare mutations found to cause congenital heart defects

Through exome sequencing and functional assays, researchers led by the Wellcome Trust Sanger Institute's Matthew Hurles uncovered rare variants linked to congenital heart defects in humans.

As the researchers reported in the American Journal of Human Genetics, they performed whole-exome sequencing of a dozen parent-offspring trios as well as of more than 110 unrelated individuals. From this, they identified a number of rare variants within the highly conserved NR2F2 gene associated with certain heart defects called atrioventricular septal defects. NR2F2, they further reported, is expressed in the developing heart, and the variants affect NR2F2 activity.

"Taken together, these data support our hypothesis that rare and private variants in NR2F2 probably contribute to AVSDs and other CHDs during human development," Hurles and his colleagues write.

According to the researchers, atrioventricular septal defects account for about 4 percent to 5 percent of all congenital heart defects, and occur in 0.3 to 0.4 of 1,000 live births.

Read the full, original story: Rare Variants Linked to Congenital Heart Defects in Humans