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Study finds link between congenital heart disease and neurodevelopmental disorders

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Researchers from Columbia University Medical Center (CUMC), NewYork-Presbyterian, the Pediatric Cardiac Genetics Consortium, and the Pediatric Heart Network have found a number of genetic mutations that explain why many children with congenital heart disease also have other significant health challenges, including neurodevelopmental disorders and other congenital problems.

The study was published in the December 3rd online edition of *Science*.

About 10 percent of babies with congenital heart disease--which affects the development, structure, and function of the heart--are later diagnosed with neurodevelopmental disorders such as learning disabilities and attention-deficit hyperactivity disorder. As many as 13 percent of children with severe forms of congenital heart disease also have other types of birth defects.

One possible explanation is that children with congenital heart disease may be deprived of sufficient blood and oxygen flow during critical moments in brain development. The investigators explored the possibility that there is a more fundamental, genetic reason behind why neurodevelopmental disorders and congenital heart disease often occur together.

"Using the sophisticated tools of genomic sequencing, we can now look deep within the genome and determine if conditions are genetically linked," said Wendy Chung, MD, PhD, the Kennedy Family Associate Professor of Pediatrics and Medicine, Director of the Clinical Genetics program at CUMC and a Clinical Geneticist at NewYork-Presbyterian/Columbia. "If such a linkage exists, then we can predict the risk that children with congenital heart disease will develop a neurodevelopmental disorder, allowing interventions to be put in place while the brain is still growing and developing. This kind of highly targeted treatment is the goal of an emerging field known as precision medicine."

In this study, the researchers used genetic information from 1,213 children with congenital heart disease and their parents to analyze the 4,000+ genes that are active during heart development. They then compared this information with previously available sequence data from 900 families not affected by congenital heart disease, to identify genetic mutations.

The study revealed that many of the children with congenital heart disease had spontaneous mutations in heart development genes. Furthermore, a single genetic mutation was responsible for about 20 percent of cases of severe congenital heart disease accompanied by neurodevelopmental disorders and/or other congenital problems. These same genes were mutated in only 2 percent of children with congenital heart disease alone, suggesting that the co-occurrence of neurodevelopmental and congenital problems has genetic causes.

The researchers also discovered that the mutations in the children born with a combination of heart, brain, and other congenital disorders occurred in a subset of genes that act like conductors, orchestrating the formation and function of organs.

"As a clinical geneticist who sees children with a variety of conditions, I find it encouraging when a study has direct and immediate benefits for patients and their families," said Dr. Chung. "We are seeing genomic sequencing move out of the realm of research and into the clinic as a diagnostic tool, bringing with it the power to predict the risk of many kinds of conditions. This is truly precision medicine at its best."

Source:
Columbia University Medical Center
