Heart disease, neurodevelopmental problems linked to similar genes

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An NIH-funded study led by scientists at Harvard Medical School who are part of the Pediatric Cardiovascular Genetics Consortium have found a link for a set of gene mutations in the development of congenital heart disease and various neurodevelopmental abnormalities in children, including cognitive, motor, social, and language impairments.

“We’re homing in on a set of genes that have multiple roles in different tissues during development: heart tissue, brain tissue, other developing organs, limb tissue,” said co-lead author Jason Homsy, M.D., a Harvard Medical School fellow. “Our study shows a common genetic link for the development of these diseases.”

The investigators utilized whole exome sequencing from more than 1,200 children and their parents to show that children with both congenital heart disease and neurodevelopmental delays share certain genetic mutations that thwart the normal development of both the heart and the brain.

“The risk of developing neurodevelopmental disabilities is so high when these particular gene mutations are present that we might consider testing for them in all patients with congenital heart disease,” explained co-author Jonathan Kaltman, M.D., a study investigator and program administrator of the National Heart, Lung, and Blood Institute’s (NHLBI) Bench to Bassinet Program, which funded the current study.

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