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Genetic Spectrum Disorders on Rodent Samples and its Impacts on Cardiac Circulations

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DESCRIPTION

Autism Spectrum Disorder (ASD) is a neurodevelopmental condition that affects social communication and behavior. ASD is also associated with various medical comorbidities, including Congenital Heart Disease (CHD), which is present in about 10% of ASD cases. CHD is a group of structural defects in the heart that impair its function and can cause life-threatening complications. The link between ASD and CHD is not fully understood, but it may involve common genetic and environmental factors that affect both the brain and the heart development. There is a strong functional and genetic connection between Autism spectrum disorder and congenital heart disease. ASD is a common (>1%) and diverse Neuro-Developmental Disorder (NDD) that mainly affects behavior, but also has other comorbidities, including cardiac ones. These cardiac comorbidities can be present at birth or develop later.

To investigate the cardiac phenotype of ASD, researchers from Canada used high-frequency ultrasound imaging to examine nine mouse models that carry mutations in genes related to ASD. These genes are Arid1b, Chd8, 16p11.2, Sgsh, Shank3, Fmr1, and Vps13b. The researchers compared the cardiac structure and function of these mutant mice with their Wild-type Littermates (WTs), which served as controls. They measured various parameters, such as heart rate, aorta diameter, left ventricular wall thickness and thickening, left ventricular chamber diameter and fractional shortening, stroke volume and cardiac output, mitral inflow peak E and A velocity ratio, and ascending aorta velocity time integral.

The results showed that the mutant mice had subtle heterogeneous cardiac abnormalities compared to WTs. Some of these abnormalities were similar to those observed in human ASD patients with CHD, such as reduced left ventricular wall thickness and thickening, increased left ventricular chamber diameter and fractional shortening, and altered mitral inflow peak E and A velocity ratio. However, the researchers also found more differences among the mutant groups than between the mutant groups and WTs, indicating a high degree of variability in the cardiac phenotype of ASD. Moreover, the type of cardiac abnormalities differed depending on the gene mutation. For example, 16p11.2 deletion mice had reduced heart rate and aorta diameter, Fmr1 knockout mice had increased heart rate and aorta diameter, Arid1b heterozygous mice had reduced left ventricular wall thickness and thickening, Sgsh heterozygous mice had increased left ventricular chamber diameter and fractional shortening, and Shank3 exon 4-9 deletion mice had altered mitral inflow peak E and A velocity ratio.

The researchers concluded that their study provides a comprehensive characterization of the cardiac phenotype of nine genetic mouse models of ASD. They suggested that their findings reflect the clinical heterogeneity of ASD and its cardiac comorbidities. They also proposed that the type of cardiac abnormalities may reveal common underlying mechanisms that affect both the brain and the heart development in ASD. For example, reduced left ventricular wall thickness and thickening may indicate impaired cardiomyocytes proliferation or differentiation, increased left ventricular chamber diameter and fractional shortening may indicate altered cardiac contractility or relaxation, and altered mitral inflow peak E and A velocity ratio may indicate impaired diastolic function or filling pressure. The researchers emphasized that understanding the cardiac abnormalities in ASD is important for clinical management, as even subtle or non-lethal defects can have an impact on normal development and quality of life. ASD-related genetic anomalies are linked with a range of cardiac problems. The variability that characterizes ASD in other traits is mirrored here, with more variations observed between mutant groups than between WTs. The changes were modest in size but important, and more model exploration is required to verify the observed patterns.

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