

Impact of Copy Number Variants and Genetic Defects on the Outcome of Neonates with Congenital Heart Disease

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BACKGROUND: Congenital heart disease (CHD) affects approximately 1% of newborns. CHD likely results from interactions between genetic and non-genetic factors. Chromosome microarray testing suggests that copy number variants (CNVs) contribute to 5-15% of cases of CHD, and CNVs have been correlated with adverse outcomes in children with CHD. There is a rising emphasis on the use of quality metrics to evaluate congenital heart programs, thus understanding if CNVs affect clinical outcomes has increasing importance.

METHODS: A single institution, retrospective study was performed to evaluate the effect CNVs have on clinical outcome in neonates who underwent congenital heart surgery. The patients were assigned Society of Thoracic Surgeons - European Association for Cardio-Thoracic Surgery Congenital Heart Surgery Mortality Categories to allow clinical outcomes to be assessed in relation to the complexity of the operation performed. Two sample t-tests or ANOVA tests and Chi-square or Fisher's Exact test were utilized for statistical evaluation of continuous and categorical data sets, respectively. Logistic regression was used to evaluate the association of CNVs with complications following surgery to adjust for multiple confounding risk factors.

RESULTS: 142 CHD patients underwent cardiac surgery within the neonatal period at the University of Iowa Stead Family Children's hospital between 2009 and 2015. Genetic testing was normal in 63 patients, abnormal in 55 patients, and not performed in 24 patients. No statistically significant differences in length of stay or intubation or in prevalence of renal, gastrointestinal, or neurological dysfunction between the groups were identified. Patients with genetic testing were subdivided into high, moderate, and low complexity case groups. No statistically significant differences in outcomes were found after accounting for case complexity.

CONCLUSION: CNVs do not appear to affect clinical outcome following congenital heart surgery in the neonatal period, regardless of case complexity. This should be considered when counseling patients prior to surgical repair.