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Congenital Heart Disease

UTILITY OF GENETIC TESTING IN INFANTS WITH ISOLATED CONGENITAL HEART DISEASE

Poster Contributions
Poster Hall, Hall C
Saturday, March 18, 2017, 3:45 p.m.-4:30 p.m.

Session Title: Congenital Heart Disease: New Diagnostic Approaches in Congenital Heart Disease
Abstract Category: 10. Congenital Heart Disease: Pediatric
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Background: Recent advances in technology, and a greater demonstration of the genetic contributors to congenital heart disease (CHD), have resulted in an expansion of genetic screening among infants with CHD. However, the utility of this screening strategy has not been well studied. We aimed to determine the incidence of pathogenic genetic findings in isolated vs non-isolated CHD answering the question "whom should we test, and which test should we send?

Methods: A retrospective review analyzing infants less than 1 year of age with CHD admitted to our cardiac ICU (January 2010 -December 2014) was performed. Patients who had renal/head ultrasounds and genetic testing (karyotype, FISH, or chromosomal microarray (CMA)) were divided into high and low risk groups. The high risk group included those with additional malformations, family history (FH) of CHD, or CHD known to be of high risk for genetic syndromes (i.e. conotruncal heart disease). Patients with isolated CHD were considered low risk.

Results: Of 446 patients who met study criteria, 278 were considered high risk, while 168 were low risk. A pathogenic finding was identified in 134 of 278 (48%) high risk patients. Only 5 of 168 (2.9%) low risk patients were found to have a pathogenic finding (negative predictive value (NPV) =97%). Of patients who had a karyotype performed, 96 (54.8%) of the high risk patients (n=176) had abnormal karyotype versus none of the 76 low risk patients (NPV=100%). In patients who had CMA, 29 of 157 (18.5%) high risk patients had pathogenic findings vs 5 of 145 (3.4%) low risk patients (NPV=96.5%). The 5 abnormalities found in low risk patients were not known genetic syndromes and did not affect management in infancy.

Conclusions: Analysis of one of the largest CHD populations using modern genetic testing methods, we found a high rate of pathogenic genetic abnormalities in patients with CHD and additional malformations, positive FH, or CHD subtypes associated with genetic syndromes. Thus, karyotype and CMA testing of these high risk infants appears to be of high yield. We found a low rate of pathogenic findings in isolated CHD suggesting that testing during early infancy may not be indicated, and required only if additional concerns arise.