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Congenital Cardiology Solutions

RISK OF CONGENITAL HEART DISEASE IN RELATIVES OF PROBANDS WITH CONOTRUNCAL CARDIAC DEFECTS: AN EVALUATION OF 1622 FAMILIES

Moderated Poster Contributions
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Background: Current risk counseling for conotruncal cardiac defects (CTD) is based on empiric estimates from multiple studies. Sibling risk of congenital heart disease (CHD) is estimated to be 2-3% for lesions such as tetralogy of Fallot (TOF) and lower (1-2%) for transposition of the great arteries (TGA). We examined the risk of CHD in relatives of probands with CTD to assist in counseling practices.

Methods: 1,622 probands with CTD and no reported chromosomal or genetic abnormalities were recruited sequentially. A three-generation pedigree was obtained for each proband by a genetic counselor detailing the presence and type of CHD in each family member. Risks and 95% confidence intervals (CI) were calculated for subgroups of relatives based on degree of relationship and proband gender. In addition, crude odds ratios (OR) and 95% CI were used to compare risks in subgroups of relatives defined by the proband's cardiac anatomy, e.g. siblings of probands with normally related (TOF, ventricular septal defect, truncus arteriosus, interrupted aortic arch) versus malpositioned (D- and L- TGA, double outlet right ventricle) great arteries. For pairs of affected relatives, concordance rates were calculated for CTD specifically and for CHD in general.

Results: The risk for any type of CHD in relatives of probands with CTD was 3.3% in siblings (95% CI: 2.6-4.1), and 1.5% (95% CI: 1.2-2.1) in a parent, consistent with previous literature. The siblings of male probands had a higher risk (4.3%, 95% CI: 3.2-5.7) than the siblings of female probands (2.5%, 95% CI: 1.8-3.4). Risk was higher in siblings for probands with normally related great arteries as compared to probands with malpositioned great arteries (OR 2.5, 95% CI: 1.4-4.5). When the proband had normally related great arteries, 73% of the affected siblings had a concordant lesion (ie CTD). In contrast when the proband had malposition of the great vessels, only 50% of the affected siblings had a concordant CTD.

Conclusions: These data suggest that risk of CHD is higher in siblings when the proband is male or has normally related great arteries. These data contribute to our understanding of the risk for CHD in the relatives of affected individuals.