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Congenital Cardiology Solutions

PHACE SYNDROME IS COMMONLY ASSOCIATED WITH UNUSUAL AND POTENTIALLY "SILENT" SEVERE AORTIC ARCH OBSTRUCTION: THE INTERNATIONAL PHACE SYNDROME REGISTRY REVIEW

Poster Contributions
Poster Sessions, Expo North
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Session Title: Congenital Cardiology Solutions: Prenatal Diagnosis, Coronary Anomalies and More
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Authors: *Peter C. Frommelt, Michelle L. Bayer, Dawn H. Siegel, Kristen Holland, Ilona Frieden, Alfons Krol, Johannes M.P.J. Breur, Elena Pope, Orli Wargon, Beth A. Drolet, Medical College of Wisconsin, Milwaukee, WI, USA*

Background: PHACE syndrome is characterized by large infantile hemangiomas of the head and neck associated with anomalies of the brain, heart, cerebral vasculature, eyes, and chest wall. Congenital heart disease (CHD) in PHACE is common but incompletely described. We sought to better characterize CHD in PHACE using a large international registry.

Methods: The medical records of 175 subjects meeting criteria for PHACE syndrome from the PHACE Syndrome International Clinical Registry and Genetic Repository were reviewed to describe associated cardiac and aortic arch pathology.

Results: 49/175 (28%) were identified as having cardiac pathology. Coarctation/interrupted aortic arch was most common, found in 27/49 (55%). The coarctation was usually characterized by bizarre long segment transverse arch narrowing with adjacent segments of aneurysmal dilatation, and 16/27 had an associated aberrant origin of a subclavian artery so that both subclavian arteries arose distal to the obstruction. Intervention was performed in 18/27 subjects, and 14/18 procedures occurred after 1 month of age despite severe obstruction at diagnosis, with 3 requiring interposition grafts during infancy. 10/49 (20%) had a right aortic arch, and 5/10 had a vascular ring requiring surgical division. Only 1 subject had a bicuspid aortic valve (without coarctation) and none had mitral valve disease. Superior systemic venous anomalies (bilateral SVC's or retroaortic innominate vein) were also frequently seen (20%). Intracardiac pathology was less common, with 11 having a VSD (3 requiring surgical closure), 2 tetralogy of Fallot, 1 pulmonary valve stenosis, and 1 tricuspid atresia.

Conclusion: This series provides the most extensive review of PHACE syndrome cardiac pathology and confirms the high risk of significant CHD. Unusual and severe aortic arch pathology is frequent and may be difficult to appreciate clinically because of associated aberrant origin of a subclavian artery. The lack of associated intracardiac findings suggests a primary aortopathy. Cardiac and aortic arch imaging with detailed assessment of arch patency and brachiocephalic origins is required upon diagnosis in all suspected PHACE patients.