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First-degree relatives of patients with hypoplastic left heart syndrome are at higher risk for cardiac malformation

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Introduction: Hypoplastic left heart syndrome (HLHS) is a rare and severe congenital heart defect characterized by a variable degree of underdevelopment of the left-sided heart structures. A higher incidence of cardiovascular malformations (CVM) in relatives of HLHS-patients and a familial clustering of HLHS has been suggested in previous studies. However these assumptions are mostly based on epidemiologic studies, detailed echocardiographic screening in affected families to detect subclinical malformations such as bicuspid aortic valves, is rare.

Objective: This study aims to investigate patterns of familial aggregation of cardiovascular malformations in families affected with hypoplastic left heart syndrome.

Methods: First-degree relatives of HLHS patients (to date n=38) were prospectively recruited at our institution. Clinical examination, 2-D and Doppler transthoracic echocardiography were performed as well as a detailed review of the medical records.

Results: We investigated the families of 21 HLHS-patients. In three families more than one child was born with a congenital heart defect (CHD). One sibling of an HLHS-patient was also affected by HLHS, one suffered from tetralogy of Fallot and the third was born with a ventricular septal defect. Transthoracic echocardiography was performed in first-degree relatives (n=35). A cardiac malformation was found in four participants (11,4 %). Two of them had a bicuspid aortic valves (BAV), one severely dysplastic with moderate aortic regurgitation. One father was born with an atrial septal defect with suspected partial anomalous pulmonary venous drainage (PAPVD). One mother was suffering from dilatative cardiomyopathy after an episode of myocarditis. Of the examined relatives one did not have previous knowledge about the CVM. Overall the incidence of CVM was 18,4 % compared to 1 % of the average population.

Conclusion: The incidence of cardiovascular malformations was highly increased amongst first-degree relatives of HLHS-patients compared to the average population. Therefore establishing a screening program for congenital heart disease should be considered in families affected with HLHS to prevent further morbidity.