Yield of echocardiographic screening of first-degree relatives of children with bicuspid aortic valve

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Background: Bicuspid aortic valve (BAV) is the most common congenital heart defect, affecting 0.8% of new-borns. BAV is associated with valve dysfunction, as well as an increased risk of aortopathy. The prevalence of BAV in first-degree relatives of symptomatic BAV patients, is reported to be 5–10%. First-degree relatives also have an increased risk of aortic dilatation, independently of aortic valve morphology.

Purpose: The purpose of the study was to determine the prevalence of BAV, associated valve dysfunction and aortopathy in first-degree relatives to children diagnosed with BAV neonatally in a population-based study.

Methods: Between April 2016 and October 2018 all expecting parents at three major maternity centres were offered inclusion in a large-scale population study with focus on congenital heart disease (N ≥ 25,000). A total of 197 children, in 196 families, were diagnosed with BAV. All first-degree relatives, including half-siblings, were offered inclusion in the follow-up study with standardized transthoracic echocardiography. Adults were also examined with transoesophageal echocardiography. Aorta diameters were measured at the AV annulus, sinuses of Valsalva, sino-tubolare junction and in the proximal ascending aorta. Aortic dilatation in children were defined as any aortic root or ascending aorta diameter ≥ 2 standard deviations (SD) from the expected mean, calculated as z-score using formulas from the Paediatric Heart Network Echocardiogram Database. In adults, aortic dilatation was defined as aortic root and/or ascending aorta diameters indexed to body surface area (BSA) exceeding normal reference values established by the European Association of Cardiovascular Imaging.

Results: In total, 352 first-degree relatives (242 adults [35.3 years SD 5.5] and 110 children [4.5 years, SD 3.5]) were included. BAV was diagnosed in 24 relatives (6.8%). BAV could not be conclusively ruled out in 52 relatives (14.8%), who are awaiting further examination with transoesophageal echocardiography. Dilatation of the aortic root was observed in 8 adult relatives and in 17 children (7.1%). Aortic valve regurgitation was observed in 23 relatives (6.5%). In total 58 relatives (16.2%) were diagnosed with either BAV, dilated aorta, or aortic regurgitation.

Conclusion: One in fifteen first-degree relatives to children diagnosed with BAV neonatally, also had BAV, corresponding to >8 fold increase in prevalence compared with the background population. One in six relatives had BAV, dilated aorta, or aortic regurgitation. These findings are of importance for family-screening programs of BAV.