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Bicuspid Aortic Valve and Dilated Cardiomyophaty: two separate entities or common genetic link?

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Introduction: Whilst the genetic association between bicuspid aortic valve (BAV) and hypertrophic cardiomyopathy (HCM) has been reported in the literature, very little is known about the coexistence of BAV and dilated cardiomyophaty (DCM). This study investigates the prevalence of coexistent DCM in a BAV population.

Methods: Retrospective data of 196 consecutive BAV patients who attended the Cardiac Outpatients Clinics between June 2012 and October 2018, were analyzed. Data included assessment of family history of inherited cardiac conditions and congenital heart diseases, ECG, transthoracic echo and cardiac magnetic resonance imaging (CMR).

Results: Out of 196 patients, 179 patients were included since free from other associated cardiac lesions which could have caused ventricular volume overload such as ventricular septal defect and patent ductus arteriosus. Mean age was 34.6±13.5, male 128 (71%). 111 patients had isolated BAV, whilst 68 patients had BAV with aortic coarctation (CoA). 43 patients required surgical or trans-catheter procedures for aortic valve (AV) dysfunction (mean age 26.1±11.1), 37 patients had CoA repair or stent (mean age 11.6±10.4). In patients who were free from previous AV procedures (n=136), there was evidence of mild AV stenosis (peak velocity 2.1±0.9 m/s) and/or regurgitation (regurgitant fraction 9±12%). Of these, 25(19%) had evidence of left ventricular (28%), right ventricular (12%) or both left and right ventricular dilatation (60%) which could not be explained by the degree of AV dysfunction or other right sided lesions for the RV. In particular,14 patients had ascertained DCM and were under specialist follow-up. On CMR, their left ventricular end-diastolic volume was 124±24ml/m^2, right ventricular end-diastolic volume 110±25ml/m^2, left ventricle ejection fraction 55±8% and right ventricle ejection fraction 57±6%. In 11 patients DCM was suspected but not yet confirmed.

Conclusions: There is a high prevalence of left and/or right ventricular dilatation in patients with BAV which is not proportional to the degree of valve dysfunction and 8% of patients had a definitive diagnosis of DCM. This is highly suggestive of a possible genetic link between BAV and DCM, although further genetic investigations will be necessary to confirm our data.