Low Risk for Aortic Root Dilation in Ehlers-Danlos hypermobility type based on repeat Echocardiogram

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Introduction:

Hypermobile EDS (hEDS) is a connective tissue disorder characterized by joint hypermobility, chronic joint pain, soft skin, and easy bruising. Aortic root dilation (AD) has been reported in up to 33% of hEDS, necessitating life-long echocardiographic surveillance. More current studies report a lower rate of AD, and as diagnostic criteria for hEDS has recently changed, the true prevalence of AD is not well known. We aimed to determine the prevalence and risk factors for AD in patients that meet strict diagnostic criteria for hEDS.

Method:

We conducted a retrospective study of patients with hEDS from 1/2008 to 12/2016. Patients included met strict diagnostic criteria for hEDS (Levy et al. GeneReviews, NIH.gov 2016) as assessed by four experienced geneticists and/or cardiologists, and also had at least one echocardiogram. Data collected included aortic root dimension and clinical features of hEDS including Beighton score, skin abnormalities, functional GI disorders, etc. Aortic root measurements were performed by experienced cardiologists and age-appropriate normative data (Colan et al. or Devereux et al.) were used for z-score calculation. AD was defined as z-score $\geq$ 2.

Results:

A total of 194 patients met diagnostic criteria for hEDS and had at least 1 echocardiogram. AD was present in 22 (11%) of patients on initial echo, of which 14 (63%) had normalization of z-score on follow-up, while 7 had persistent AD. Those with persistent AD were more likely to have family history of AD and mitral valve prolapse, and had lower rates of chronic headaches, functional GI disorders, or POTS (all variables p-value<0.05).

Conclusion:

Patients who meet strict diagnostic criteria for hEDS have lower risk for AD than previously described. Those with AD are more likely to have family history of AD and less likely to have non-cardiac systemic manifestations of hEDS.

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