Variability of different Z-score calculations of the aortic sinus of Valsalva in Marfan syndrome and in healthy children

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Introduction: Aortic root aneurysm is one of the key diagnostic criteria of Marfan syndrome, and its rate of dilatation determines clinical course and management. 2D-echocardiographic dimensions of the sinus of Valsalva (SoV) are standardised for age, gender and body size, and are expressed as Z-scores, with values ≥2 suggestive of aortic root dilatation. The aim of this study was to assess the potential variation of different published Z-score algorithms used in clinical practice.

Methods: 23 patients with fibrillin-1 mutation-positive Marfan syndrome (according to the revised Ghent nosology) (48% female, 11.1 ±4.1 years) and 23 healthy children (56% female, 11.5 ±3.7 years) were assessed retrospectively. Echocardiographic measurements were performed using both inner edge and leading edge conventions at end-systole and end-diastole. Z-scores were calculated using published methods of Warren et al. (Halifax), Pettersen et al. (Detroit), Daubeney et al. (Wessex), Gautier et al. (Paris), and the recently published Z-score equations based on the largest cohort of children and adults (849 subjects) by Campens et al.

Results: The mean diameter of SoV was significantly larger in the Marfan cohort than in healthy children (30 ±6.3mm vs 23 ±3.4mm, p<0.01) with mean Z-scores varying from 1.6 ±1.50 (Paris) to 2.77 ±2.09 (Wessex) in the Marfan group compared to -0.95 ±1.01 (Paris) to -0.11 ±0.85 (Halifax) in the control group. There was substantial variation in the proportion of patients fulfilling criteria for aortic root dilatation (Z≥2) according to different calculations: 43% (Detroit and Paris), 57% (Halifax), 61% (Wessex), and 65% and 74% (Campens et al.). Z-scores in the control group were all less than 2, except for one value of 2.06 from the Wessex dataset.

Conclusions: The variability of Z-score results derived from the dimension of the sinus of Valsalva by different Z-score equations has important implications for the diagnosis of Marfan syndrome in clinical practice. Z-score data in children with Marfan syndrome should therefore be interpreted with caution, and further, prospective studies are required to establish the accuracy of different Z-score calculation algorithms.