

New Screening Tool for Aortic Root Dilatation in Children with Marfan Syndrome and Marfan-like Disorders

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Background

The primary role of pediatric cardiologist, consulting patients with a suspicion or diagnosis of a genetically determined connective tissue disease (such as Marfan, Ehlers-Danlos or Loeys-Dietz syndromes) is to assess whether aortic root is dilated. Patient's age, gender and body surface area influence aortic root diameter, therefore, it is not possible to establish its single normal range for entire population. Thus it is necessary to assess aortic root diameter with special nomograms and express it in z-score. Z-score calculations are time-consuming and could be troublesome, if used infrequently. This study was aimed at introducing a simple screening method for identification aortic root dilatation in children.

Methods

Study population consisted of 190 children (3 months -18 years) with the diagnosis of Marfan syndrome or Marfan-like disorders. Aortic Root ratio (ARr) was created, which was a simple quotient of aortic root diameter to patient's height (Fig.1). The value of ARr in each patient was confronted with the results obtained using three most widespread z-score calculators (based on nomograms by Gautier et al., Pettersen et al. and Cantinotti et al.). The ROC curves analysis was employed to evaluate the predictive value of ARr to identify aortic root dilatation and to determine the optimal cut-off value of ARr.

$$\frac{\text{aortic root diameter (mm)}}{\text{patient's height (cm)}} * 100$$

Fig.1. Formula for calculation ARr.

Results

Surprisingly, we revealed that three commonly used z-score calculators were not perfectly concurring with each other – in as many as 5.8% of patients the results (dilated or non-dilated aortic root) were divergent between them. The calculated optimal cut-off value of ARr was ≥ 18.7 . At that cut-off point the sensitivity of ARr ranged from 88.3% to 100% and the specificity ranged from 94% to 97.8% (depending on z-score calculator applied for comparisons). In all patients, in whom ARr at that cut-off point failed to identify aortic root dilatation, the results were divergent using different z-score calculators. At the cut-off point of ≥ 18.0 the sensitivity of 100% was achieved for all z-score calculators.

Conclusions

ARr allows for rapid and precise screening for aortic root dilatation in children. Unlike the classic analysis, ARr doesn't necessitate access to any nomograms or on-line calculators.