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Pediatric Friedreich's Ataxia: new echocardiographic data for the follow-up

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Introduction.
The main cause of mortality in Friedreich's Ataxia is cardiac disease. Strict follow-up with clinical and echocardiographic data is essential in this group of patients. Latest data in some published adult series showed the role of relative wall thickness (RWT) and its relation with an early detection of concentric remodeling despite of a normal left ventricular mass index (LVMI).
We present a retrospective description of echocardiographic measurements and their relation with demographic and genetic data.
Methods.
We analysed retrospectively a cohort of 11 pediatric patients diagnosed with Fridreich's Ataxia. Demographic, genetics (GAA triplets), clinical and echocardiographic data were described. Echocardiographic data included left ventricle diameters and RWT (M-mode), systolic function (M-mode and Simpson), and myocardial strain with speckle tracking with CMQ of QLAB® of Philips (circumferential and longitudinal analysis). Qualitative and quantitative variables were analysed, establishing a significance level when $p<0.05$. The adjustment of the linear regression was with the R2 criteria. Statistical analysis performed with StatCrunchTM.
Results.
A total of 11 patient were analysed: 8-19yo, mean age 14.3±3.55, 72% females. Nine of them had symmetric hypertrophic cardiomyopathy, increased LVMI, and preserved left ventricle ejection fraction (LVEF,%) and 2 had normal values of LVMI and borderline values for interventricular and posterior wall thickness. RWT value was ≥0.42(mean 0.61±0.12), with no difference by gender ($p<0.05$), so despite of a normal LVMI all of them showed a concentric remodeling pattern. We detected a correlation between intervertricular septum thickness and GAA expansion, not affected by gender or age, with a significant positive correlation ($P=0.02$; $R2=0.37$), but that observation was not significant for RWT values. Myocardial strain was analysed and we obtained decreased longitudinal peak-systolic values despite of a preserved LVEF.
Conclusions.
(1)Concentric remodeling was observed in all our pediatric patients because of compatible RWT values, despite of normal LVMI. (2)GAA expansion showed a positive correlation with the interventricular septum thickness, but no with RWT. (3)Global longitudinal myocardial strain showed decreased values despite of a preserved ejection fraction.