Friedreich’s Ataxia in pediatric patients: global and segmental echocardiographic assessment

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Introduction
Friedreich's Ataxia (FA) is an autosomal recessive neurodegenerative disorder resulting from deficiency of frataxin. Cardiac impairment presents hypertrophic and dilated cardiomyopathies, ventricular dysfunction and arrhythmias. Little is known in cardiac presentation in children. We hypothesized that strain echocardiography could be a potential marker for early left ventricular dysfunction despite normal left ventricular ejection fraction even in early stages in children and young adults.

Methods
Retrospective and prospective, 14 patients with FA were analyzed (ECG and echo - global LV systolic function using Simpson’s method and for regional wall motion abnormalities using advanced CMQ (QLAB Philips (r)) for the longitudinal and circumferential strain analysis).

Results
75% men, mean age when first seen in cardiology was 7 years (5-14 years).

Echocardiography: Hypertrophic cardiomyopathy was detected in 12/14 (85%), one of them hypertrophic obstructive cardiomyopathy with mild-to-moderate mitral regurgitation. All cases of hypertrophic cardiomyopathy had threshold levels or a decreased diastolic disfunction. All of cases had a normal ejection fraction using Simpson’s method (58%-79%). One with mid hypertrophy, a regional dysfunction was detected when the longitudinal strain was performed. In all cases of hypertrophy cardiomyopathy, longitudinal strain analysis showed a reduction with segmental variation that was not consistent to a particular region.

Arrhythmias: two patients with hypertrophic cardiomyopathy had asymptomatic atrial extrasystole registered in the 24h-cardiac Holter. No other arrhythmias had been detected.

ECG: we detected T wave changes in all cases with hypertrophic cardiomyopathy, deep S wave and high R wave in leads V2-V4, and incomplete right branch block in two cases. PR and QTc intervals were normal.

Treatment: all cases were under treatment with idebenone or idebenone plus vitE/CoQ10/beta blocker. Betablocker therapy was started when an obstructive hypertrophy or diastolic dysfunction were detected.

Conclusions
Children with FA and hypertrophic cardiomyopathy had an asymptomatic diastolic disfunction and segmental disfunction detected with longitudinal strain with a normal ejection fraction. Thus, segmental echo should be performed to all patients with FA.