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

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Introduction

Cardiac associations with Friedreich’s Ataxia (FRDA) are hypertrophic and dilated cardiomyopathies, ventricular dysfunction and arrhythmias.

Advanced echocardiographic assessment with Strain could be a potential marker for early left ventricular dysfunction despite normal left ventricular ejection fraction even in early stages in children.

Objectives

To determine retrospectively:
- Epidemiologic: age, onset-age, sex, clinical data and treatment
- Echocardiography: ejection fraction by M-mode and Simpson, left ventricular mass. To analyze Strain with advanced CMQ with software Philips QLAB®.
- ECG: electrocardiographic and 24h-cardiac Holter features.

Material and methods

Retrospective analysis in 14 patients with FRDA were performed.

ECG, 24h-cardiac Holter and echocardiograms of all patients with FRDA were analyzed.

We registered global left ventricular systolic function using Simpson’s method and for regional wall motion abnormalities using Philips advanced CMQ® for the longitudinal and circumferential strain analysis.

<table>
<thead>
<tr>
<th>Echocardiographic assessment</th>
<th>EF (%)</th>
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<tbody>
<tr>
<td>LV Max Systole (g)</td>
<td>48.6</td>
</tr>
<tr>
<td>SAX GCS</td>
<td>-29.4</td>
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<tr>
<td>SAX GLS</td>
<td>-24.5</td>
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<tr>
<td>GLS Max A4C</td>
<td>-25.3</td>
</tr>
<tr>
<td>GLS Max A2C</td>
<td>-24.4</td>
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<tr>
<td>GLS avg</td>
<td>-23.7</td>
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</table>

Epidemiology: 75% were boys (median age 7 years). We found data for assessment of global ventricular function in 14 patients and advances functional assessment with strain in 10 patients. No sudden death in pediatric age was registered in our center. No syncope or chest pain was registered in the clinical reports.

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 dysfunction. All cases had a normal ejection fraction using Simpson’s method (58-79%). One with mild hypertrophy, a regional dysfunction was detected when the longitudinal strain was performed. In all cases of hypertrophic cardiomyopathy, longitudinal strain analysis showed a reduction with segmental variation that was not consistent to a particular region.

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

Treatment: all cases were under treatment with idebenone or idebenone plus vitE/CoQ10/beta-blocker or ACE inhibitor.

Discussion

1. Children with FRDA and hypertrophic cardiomyopathy had an asymptomatic diastolic dysfunction and segmental dysfunction detected with longitudinal strain with a normal ejection fraction.

2. Seems to be important an advanced assessment of children with FRDA in order to detect this dysfunction early.

3. Whether there is clinical or prognosis value of any of these Strain measurements in FRDA is unknown at this time.

4. Latest studies (Martin St John Sutton et al) showed that a decreased longitudinal strain and radial strain tended to a longer GAA repeat length (p = 0.088 and 0.083 respectively). We found this trend in our patients. More patients are needed to correlate these features.