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Assessment of global & regional ventricular function in children with Friedreich's Ataxia: a single centre experience

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Introduction: Friedreich's ataxia (FA) is an autosomal recessive neurodegenerative disorder, caused by unstable GAA expansions in the FA gene encoding for the 210-amino-acid protein frataxin. Cardiac associations described are hypertrophic cardiomyopathy (HCM), dilated cardiomyopathy (DCM), ventricular dysfunction and arrhythmias. There is dearth of literature in paediatrics of cardiac presentation and progression in FA.
Methods: 13 year retrospective analysis of data in a single centre; clinical features, electrocardiographs (ECG) and echocardiograms of all patients with FA looking for patterns of presentation and progression of cardiac disease.
Results: 21 patients with confirmed diagnosis of FA were seen (10 male and 11 female). Analysis was available in 18 patients (insufficient data in 2 patients) for assessment of global ventricular function, and advanced (synchronicity & strain) functional assessment in 10 of these. Median age of presentation to cardiology was 11 years (5-15 years).
ECG abnormalities in 12 (63%) with isolated T wave inversion in lateral leads in 8(42%), left ventricular hypertrophy with strain pattern in 4, normal in 6 (no data in 1). None had palpitations/syncope.
Echocardiograms assessed for left ventricular mass (107–209G), global and regional function. 15 (83%) had concentric HCM; of these 13 had normal global systolic function, three had diastolic dysfunction. One had DCM, and one (5%) had hypertrophic obstructive cardiomyopathy (HOCM), and no hypertrophy in two.
Synchronicity parameters were within normal limits in all 10 patients we analysed; all segment delay varied from 45-165ms (SD: 6-45ms). Longitudinal strain (ε) showed a reduction in all patients (average ε range was -9.1% to -20.6%) with segmental variation that was not consistent to a particular region.
Nine patients were on idebenone; three were on additional Vitamin E and Coenzyme q10; and one on beta-blocker (HOCM). There were no arrhythmias noted in our case series.
Conclusion: Children with FA have some evidence of cardiac dysfunction, but are asymptomatic. The dominant pattern is concentric LVH, but other patterns are also present, and there are regional changes even when global function is normal. Regular, serial assessment is essential in this group preferably with advanced analysis looking for progression.