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Assessment of cardiac 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 with FA are hypertrophic (HCM) and dilated (DCM) cardiomyopathies, ventricular dysfunction and arrhythmias. Presentation and Cardiac abnormalities in the paediatric population with FA are not well described. We aim to look at these.

Material and Methods: 12 year retrospective analysis of data in a single centre; clinical features, electrocardiographs (ECG) and echocardiograms of all patients with FA analysed looking for patterns of presentation and progression of cardiac disease.

Results: 21 patients with confirmed diagnosis of FA were seen (8 male and 13 female). 19 patients analysed (insufficient data in 2 patients). Median age of presentation to cardiology was 11 years (5-15 years). Referrals were from paediatricians/neurologists, 10 with confirmed diagnosis at referral, 7 with neurological signs and two with heart murmurs. All had progressive neurological signs, five were wheel chair bound. Three had kyphoscoliosis with two needing spinal surgery. ECG abnormalities at presentation were noted in all with isolated T changes (100%); Left ventricular hypertrophy (LVH) in 13 (66%). None had palpitations/syncope or arrhythmias. Echocardiograms assessed for left ventricular mass, global and regional function. At presentation, 16 (84%) had concentric HCM; of these 11 had normal global systolic function, with additional diastolic dysfunction in three. Two (10%) had DCM, and one (5%) had hypertrophic obstructive cardiomyopathy (HOCM). Regional wall motion abnormalities were seen in all patients. 16 patients had serial scans, six (31%) had some improvement of cardiac lesion; these were also on Idebenone treatment. Vitamin E and Coenzyme q10 was used in three; and one patient with HOCM on beta-blocker. There was progression of LVH and or dilatation in 3 (16%). All are alive; nine have ongoing reviews and 10 transferred to adults.

Conclusions: Concentric hypertrophied cardiomyopathy is the most common cardiac abnormality with specific ECG changes seen in all of them. Regional wall motion abnormalities were seen even where global function was normal. This highlights the importance of undertaking detailed ventricular analysis for assessment of both global and regional function.