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High Prevalence of Myocardial Fibrosis and Left Ventricular Hypertrophy in Pediatric Friedreich's Ataxia: a Cardiac Magnetic Resonance Imaging Study

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Background: Friedreich's Ataxia (FA), an autosomal recessive neurodegenerative disorder, is the most common cause of ataxia in the US (1 in 40,000 Caucasians). Cardiomyopathy is present in approximately 92% of patients, and is the leading cause of death.

Purpose: To determine the prevalence of left ventricular hypertrophy (LVH) and myocardial fibrosis in pediatric patients with FA.

Methods: Forty eight pediatric patients with FA were recruited for a double blind placebo controlled National Institutes of Health sponsored study of Idebenone. Prior to the investigational treatment, all of the patients were offered a cardiac MRI cine study with the option to receive contrast. All patients were scanned on a Siemens 1.5 Tesla Avanto magnet using an 8 element phased array coil. Cine imaging was performed with steady state free precession (SSFP). Delayed enhancement (DE) imaging was performed using a single-shot phase-sensitive inversion-recovery SSFP sequence, 10 minutes post-administration of 0.15 mmol/kg gadolinium-DTPA. Additional higher resolution DE images were obtained in long and short axis views using a phase-sensitive inversion recovery gradient echo sequence to confirm abnormal regions. Left Ventricular Ejection Fraction (LVEF) and myocardial mass were measured by planimetry, and DE was determined by a consensus of two cardiologists.

Results: 45/48 patients completed the cine MRI, and 36/45 completed the gadolinium DE imaging. There were no complications. There were 24 males and 21 females. The average LVEF was 64.1% +/-7.1, 6/45 (13%) had an abnormal LVEF of less than 55%. The average myocardial mass in diastole was 73.7 +/- 19.6 and in systole was 75.2 +/- 20. The prevalence of LVH was 27% (12/45 patients) as defined by a myocardial mass >80g/m². The prevalence of atypical DE was 50% (18/36 patients). The DE pattern was similar to that seen in myocarditis and was predominately located in the mid wall, patchy, and did not follow the distribution of coronary arteries. Conclusions: DE abnormalities are more common than either LVH, or decreased LVEF. This abnormal DE pattern likely represents myocardial fibrosis. Thus, myocardial fibrosis in these patients may be an early indicator of disease progression, and LVH may be secondary to the myocardial fibrosis.