

MP5-9

Clinical presentation and survival of childhood hypertrophic cardiomyopathy associated with Friedreich ataxia: a national cohort study.

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Background:

The clinical spectrum of disease and survival in childhood hypertrophic cardiomyopathy (HCM) associated with Friedreich ataxia (FA) is poorly described. This study describes the clinical characteristics and outcomes of patients with FA associated HCM over four decades in a well-characterised United Kingdom (UK) cohort.

Methods

Demographic and clinical data for all children diagnosed with HCM secondary to FA between 1980 and 2017 were retrospectively collected.

Results

74 patients with FA (39 male [53%]) met diagnostic criteria for HCM at a mean age 10.6 years (+3, range 4.6-17.1). In 28 patients (38%) the diagnosis of HCM preceded that of FA. At the time of diagnosis: 24 patients (32%) were symptomatic (chest pain n=10; palpitations n=6; pre-syncope/syncope n=7; NYHA >2 n=12); 62 (84%) had concentric left ventricular hypertrophy with a mean LV maximal wall thickness 12mm (+/- 2.6, range 8-19mm); 1 (1.4%) had obstructive disease (LV outflow tract gradient >30mmHg); and 4 (5.4%) patients had impaired systolic function (EF < 50%). Over a mean follow up of 5.2 years (+/-4.7, range 0.5-28.7), 8 patients (12%) had documented supraventricular arrhythmias (Atrial flutter n=3, Atrial ectopic tachycardia n=1, Atrial fibrillation n=4, re-entry tachycardia n=1), 3 (4%) had ventricular arrhythmias (non-sustained ventricular tachycardia n=3) and 1 (1.4%) had conduction disease. Freedom from supraventricular arrhythmias was 98.6 (95% CI 90.7-99.8) at 1 year and 74.7% (95% CI 49.4-88.7) at 10 years. 39 patients (53%) were started on medications for cardiac symptoms (B-blockers n=16, Calcium channel blocker n=7, heart-failure medications n=5, Anti-arrhythmics n= 4, other n= 7), 1 underwent a cardiac transplantation (aged 4 yrs.), 1 suffered a transient ischaemic attack (aged 13 yrs.) and 4 patients (5.4%) died (congestive cardiac failure n=1, atrial arrhythmia-related n=2, non-cardiac n=2). There were no sudden cardiac deaths. Overall mortality rate was 1.35 (95% CI 0.61-3.01)/100 patient years follow up.

Conclusions

This national study of FA-associated childhood HCM is the largest reported and describes a symptomatic cohort of patients with a high prevalence of childhood atrial arrhythmias and early progression to end stage disease. Overall mortality is similar to that reported in non-syndromic childhood HCM but no patients died suddenly.