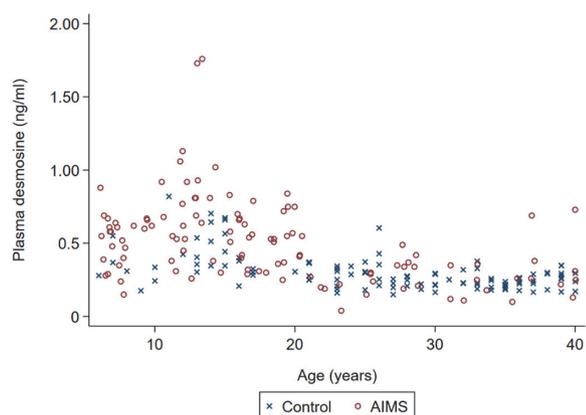


Abstract 5 Figure 1



Abstract 5 Figure 2

pDES compared to controls before the age of 20 ( $p=0.01$ ) but in adulthood, there was no difference (figure 2).

**Conclusion** Elastin turnover is highly dynamic before early adulthood, and peaks in adolescence and is exaggerated in MFS, suggesting that this period of growth is critical in developing aortopathy.

**Conflict of Interest** None

## 6 CLINICAL PRESENTATION AND OUTCOMES OF CHILDHOOD HYPERTROPHIC CARDIOMYOPATHY ASSOCIATED WITH FRIEDREICH'S ATAXIA: A NATIONAL COHORT STUDY

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**Introduction** Hypertrophic cardiomyopathy (HCM) is a common manifestation and important predictor of morbidity and mortality in children with Friedreich's ataxia (FA). HCM is present in up to 85% of FA patients however despite this, the clinical spectrum and phenotype is poorly described in children. This has important implications for long-term follow-up and management of children with this disease.

The aim of this study was to describe the clinical characteristics and outcomes in a well-characterised longitudinal cohort of children with FA-HCM in the United Kingdom (UK) over four decades.

**Methods** Demographic and clinical data for all children (<18 years) diagnosed with FA-HCM between 1980-2017 were retrospectively collected from 11 out of 13 UK paediatric cardiology centres.

**Results** 78 patients with FA-HCM were identified (male  $n=45$ , 58%) with a mean age at baseline evaluation of 10.9 years ( $\pm 3.1$ ). HCM diagnosis was within one year of cardiac referral in 29 (69.4%), but preceded the diagnosis of FA in 4 (5.3%). At baseline, 65 (90.3%) had concentric left ventricular (LV) hypertrophy with a mean maximal left ventricular wall thickness 12.8mm ( $\pm 2.6$ , range 8-19mm). Six patients (12.5%,  $n=49$ ) had LV systolic impairment. Over a median follow up of 5.1 years (IQR 2.4-7.3), eight (10.5%) had documented supraventricular arrhythmias [atrial fibrillation ( $n=3$ ), atrial flutter ( $n=1$ ), atrial ectopic tachycardia ( $n=1$ ) and unspecified supraventricular tachycardia ( $n=3$ )] and four (7.8%) had non-sustained ventricular tachycardia. There were no differences in baseline clinical characteristics between patients with and without atrial arrhythmias, but a higher proportion had impaired systolic function at last clinical review ( $n=3$ , 37.5% vs  $n=1$ , 1.4%,  $p$  value <0.001). Forty (56.3%) were started on medications for cardiac symptoms, one underwent cardiac transplantation (aged 4 years, prior to diagnosis of FA) and 8 patients (10.6%) died [atrial-arrhythmia related ( $n=2$ ); heart-failure related ( $n=1$ ); non-cardiac ( $n=2$ ); or unknown cause ( $n=3$ )]. There were no sudden cardiac deaths. Freedom from death or transplantation at 10 years was 80.8% (95% CI 62.5-90.8). No demographic or baseline clinical characteristics, including MLVWT at baseline, predicted transplant-free survival on univariable analysis (table 1).

**Conclusions** This national study of childhood FA-HCM is the largest cohort reported to date and describes a high prevalence of 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.

**Conflict of Interest** None

**Abstract 6 Table 1** Univariate cox regression analysis for predictors of outcomes

Clinical predictor	Mortality or cardiac transplantation		Atrial arrhythmia	
	Hazard ratio (95% CI)	P Value	Hazard ratio (95% CI)	P Value
Male gender	0.495 (0.11-2.21)	0.357	0.483 (0.11 - 2.11)	0.324
Any symptoms at baseline	1.053 (0.19-5.79)	0.953	0.904 (0.16 - 4.95)	0.907
Heart failure symptoms	0.888 (0.11 - 7.41)	0.912	0.862 (0.11 - 7.02)	0.887
Increasing LVMWT	0.735 (0.49 - 1.10)	0.089	0.946 (0.72 - 1.24)	0.687
Increasing LVOT gradient	0.956 (0.80 - 1.14)	0.516	0.936 (0.75 - 1.17)	0.418
Impaired LV systolic function	3.162 (0.52 - 19.11)	0.237	1.360 (0.15 - 12.20)	0.790
Impaired LV diastolic function	NA	NA	1.620 (0.17 - 15.75)	0.690
Atrial arrhythmia	1.834 (0.37-9.12)	0.482	NA	NA

CI= confidence interval, LVMWT = left ventricular maximal wall thickness, LVOT =left ventricular outflow tract