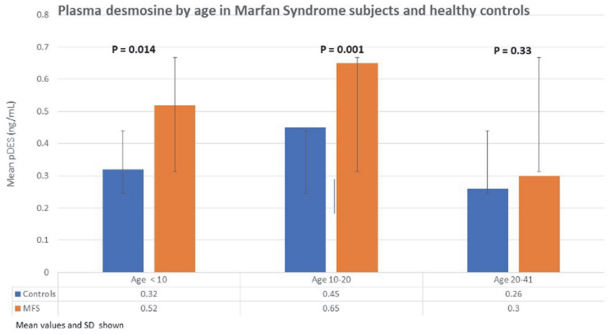
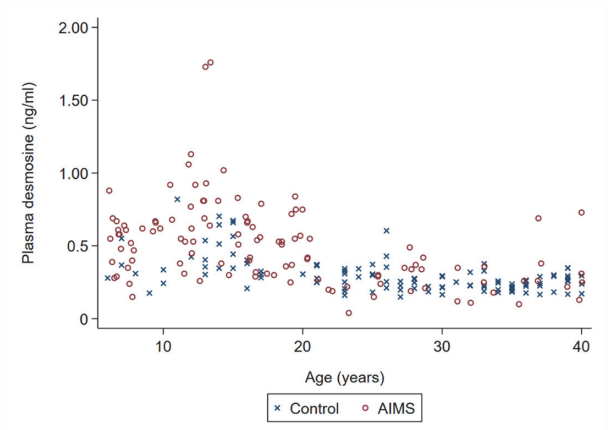


Abstract 5 Figure 1



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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10.1136/heartjnl-2021-BCS.6

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 (± 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 (± 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