

PW1-6

Aetiology, Presentation, and Outcomes of Hypertrophic Cardiomyopathy in Childhood in Wales

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Background: Hypertrophic cardiomyopathy (HCM) is commonly caused by a mutation in a sarcomeric protein, or secondary to a neuromuscular disorder, malformation syndrome or inborn error of metabolism. The purpose of this study was to establish the causes, management and outcomes of HCM in the Welsh paediatric population.

Methods: Patients diagnosed with HCM in Wales between 1984 and 2011 were included in the review.

Results: Of 58 patients diagnosed with HCM, 17 (29.3%) cases were familial while 41 cases (70.7%) were secondary to another cause. 21 of these secondary cases (36.2%) resolved with treatment while the remaining did not. The most common identification of familial HCM was through genetic testing of patients with a family history, with the majority (64.7%) being diagnosed over 15 years old. While secondary HCM was most commonly identified by neonatal screening with 80.5% being diagnosed during infancy. All patients with non-resolving secondary HCM had an abnormal baseline ECG, abnormal echocardiogram or both. Of the patients with familial HCM three (17.6%) of genotype positive patients had completely normal investigations. Five patients (29.4%) with familial HCM received an ICD to prevent sudden cardiac death (SCD). The four (6.9%) patients from the secondary HCM group died; only two of these deaths were cardiac related, one as a result of heart failure, the other due to arrhythmic SCD.

Conclusions: Despite the eminent risk of SCD in patients with familial HCM, no patients in this cohort died as a result of this and most remained asymptomatic. The prognosis for secondary HCM was highly variable depending upon the cause. Those born to diabetic mothers had an excellent prognosis with all cases resolving in time; in contrast those secondary to metabolic diseases had much poorer outcomes.