

Yield of clinical screening for hypertrophic cardiomyopathy in child first-degree relatives: evidence for a change in paradigm

Norrish G. (1), Jager J. (1), Field E. (1), Quinn E. (3), Fell H. (1), Lord E. (1), Cervi E. (1), Elliott P.M. (2,3), Kaski J.P. (1,2)

(1) Centre for Inherited Cardiovascular Diseases, Great Ormond Street Hospital, London UK; (2) Institute of Cardiovascular Sciences, University College London, UK; (3) St Bartholomew's Center for Inherited Cardiovascular Disease, St Bartholomew's Hospital, London, UK

Background:

Hypertrophic Cardiomyopathy (HCM) is a heritable myocardial disease with age related penetrance. Current guidelines recommend clinical screening of relatives from the age of 10 years onwards but the clinical value of this approach has not been systematically evaluated

Methods and results:

Anonymized, clinical data were collected from children referred for family screening between 1994-2017 following diagnosis of HCM in a first-degree relative. 1198 consecutive children (aged ≤ 18 years) from 594 families underwent serial evaluation [median 3.5 years (IQR, 1.2-7)]; 32 individuals met diagnostic criteria at baseline (median maximal LV wall thickness (MLVWT) 13mm (IQR, 8-21mm)) and 25 additional patients developed HCM over 4.6 years (IQR 2.8-7.1 years). Median age at first diagnosis was 10 years (IQR 4-13) and 41 (72%) were 12 years or younger at diagnosis (*figure*). Median age of affected patients at last follow up was 14 years (IQR 9.5-18.2). A family history of childhood HCM was more common in those patients diagnosed with HCM (n=32, 56%, VS n=257, 23% P <0.001). 18 patients (32%) were started on medication for symptoms, 2 (4%) underwent a septal myectomy, 14 (25%) received an implantable cardioverter defibrillator, 1 underwent cardiac transplantation, 2 had a resuscitated cardiac arrest and 1 died secondary to a cerebrovascular accident.

Conclusions:

Almost 5% of first-degree child relatives from 8% of families undergoing screening meet diagnostic criteria for HCM at first or subsequent evaluations, with the majority presenting as pre-adolescents. The phenotype of familial HCM in childhood is varied and includes severe disease, suggesting that clinical screening should commence at a younger age

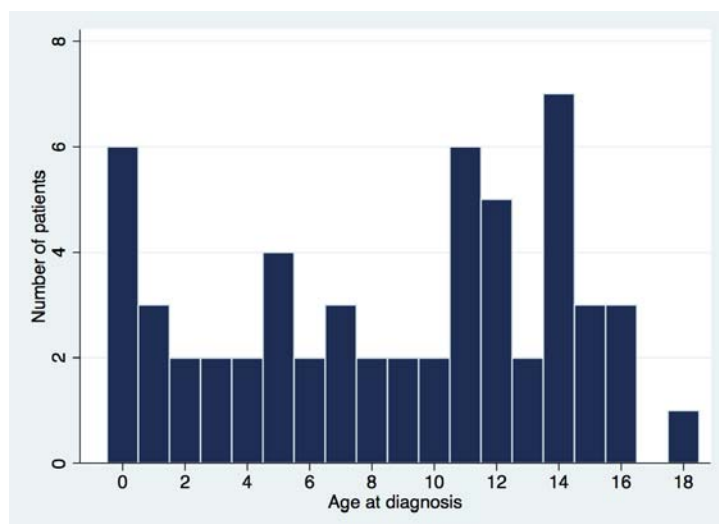


Figure: Age of patients at diagnosis [≤ 12 years (72% n=41), ≤ 10 years (52%, n=30)]