Comparison of echocardiography and ECG for detection of abnormal phenotype in childhood mutation carriers for familial hypertrophic cardiomyopathy

Allahyari, P., Östman-Smith, I.
Division of Paediatric Cardiology, Institute of Clinical Sciences, Queen Silvia Children’s Hospital, Gothenburg, Sweden

BACKGROUND Studies from tertiary centres specialised in hypertrophic cardiomyopathy (HCM) have claimed that frequency of abnormal ECG in affected adults range from 75-97% but data from geographically based cohorts and childhood patients are lacking. We have therefore studied a geographical cohort of childhood patients positive for a mutation causing familial HCM.

METHODS 36 mutation carriers, age 0.2-18 years (median age 10yrs), identified from a systematic screening project in the West Götaland region in Sweden, were compared with age and gender-matched normal controls. The parameters compared were measures that have been suggested useful for screening for early detection of abnormal phenotype. Firstly, M-mode indices of relative wall thickness and contractility from long-axis M-mode: septum-to-cavity ratio (sepcavr), left ventricular wall-to-cavity ratio (lvcavr), posterior wall systolic wall-to-cavity ratio (syscavr) and fractional shortening (FS). Secondly, electrocardiographic measures such as QRS-amplitude sum in limb-leads (LLQRSS), twelve-lead QRS-amplitude sum (TwQRSS), twelve-lead amplitude-duration product (TwLProd). In addition the ECG-risk score was quantified. Data were compared with Mann-Witney U-test.

RESULTS Comparing mutation carriers with controls neither LLQRSS (median values 6.5 and 6.7mV respectively), TwQRSS (20.7 versus 19.3mV) or TwLProd (1.64 versus 1.68mV.s) were significantly different. The most sensitive measure cut-off was TwQRSS >2.4mV where 28% of mutations carriers were positive, but there were also 16% false positives among normals. Pathological Q-waves were present in 43% of mutation carriers but only in 3% of normals so useful. ECG-risk score was significantly higher in mutation carriers (p=0.0002), and 19% of mutation carriers had a high-risk score of 6 or above, versus none among controls, where the highest value was 3. Sepcav, lvcavr, syscavr and FS are all significantly increased in mutation carriers (p<0.0001 in all). A cut-off of ≥0.26 has a sensitivity of mutation carriage of 75%, as has a syscavr ≥0.55, lvcavr ≥0.22 has a sensitivity of 47%, all with no false positives in normals. FS ≥42% has a sensitivity of 64% and no false positives.

CONCLUSIONS 57% of childhood mutation carriers for familial HCM have normal ECGs, whereas M-mode indices of relative septal thickness, and radial systolic contractility, are abnormal in 75% of mutation carriers already in childhood.