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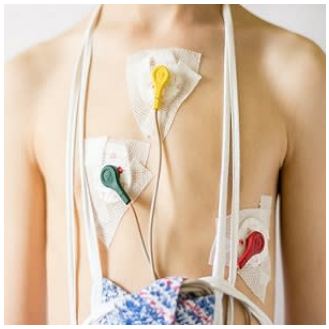
Family screening for hypertrophic cardiomyopathy



The American College of Cardiology/American Heart Association (ACC/AHA) guidelines recommend that family screening for hypertrophic cardiomyopathy (HCM) should start at the age of 12 years. Screening earlier than this is only recommended in cases with an early growth spurt, family history of sudden cardiac death and prior to competitive sports participation. The European Society of Cardiology (ESC) guidelines recommend that family screening should begin at the age of 10, and earlier screening should be considered in families with malignant, early onset of disease, presence of cardiac symptoms or engagement in activities that demand physical activity. However, there is some debate as to whether these screening guidelines are accurate, or is there a risk of missing earlier onset disease if the minimum age is set at 10 to 12 years?

A study was conducted to evaluate if the current screening guidelines miss early-onset disease, which, in turn, could have an impact on timely interventions. Study participants comprised of 524 children who underwent echocardiography prior to 18 years of age as part of family screening for primary HCM. Children who were first degree relatives of HCM probands were screened independent of age. Screening involved a clinic visit, an ECG, an echocardiogram, and genetic counselling.

Findings of the study showed that 9.9% of the children screened for HCM were already phenotype-positive at first evaluation. 52.5% of children with clinical HCM became phenotype positive before 10 years of age and 41% major cardiovascular events (MACE) occurred in children before 10 years of age. Findings suggest that many children already had a well-established phenotype by the time they were screened and demonstrated a rapid progression on follow-up. Nearly 31% children with early onset HCM did not meet eligibility criteria for early screening and would have missed detection if the guidelines were adhered to.

It is important to note that every first-degree relative from an HCM family has a 50% chance of inheriting the disease, regardless of their age. And yet, the guidelines for family screening are so designed that they underrepresent children and pre-adolescents, even though findings from this study as well as a previous report by Norrish et al. suggest that the prevalence of disease in children and pre-adolescents is not lower than in adolescents.

Overall, findings from this study suggest that clinical and genetic screening for family members should not be delayed until 10 or 12 years of age. While there may be costs associated with early screening, missed diagnosis and the preventable loss of young life can have a much higher emotional and financial cost to families and to the society in general.

Source: European Journal of Cardiology
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