

Hypertrophic cardiomyopathy – screening and etiology detection

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KEYWORDS: Anderson-Fabry disease, ATTR amyloidosis, genetic testing, hypertrophic cardiomyopathy, left ventricular hypertrophy.

CITATION: *Cardiol Croat.* 2019;14(3-4):66. | <https://doi.org/10.15836/ccar2019.66>

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Left ventricular hypertrophy is an adopted response to physiological and pathological stress, while hypertrophic cardiomyopathy (HCM) is defined by the presence of increased left ventricular wall thickness that is not solely explained by abnormal loading conditions. Determination of the etiology, pathophysiology and disease severity is important for the management of patients with HCM.¹⁻⁴ According to latest European Cardiology Society Guidelines for HCM², etiology in adults are 60% an autosomal dominant trait caused by mutations in cardiac sarcomere protein genes, 5-10% caused by other genetic disorders including inherited metabolic and neuromuscular diseases, chromosome abnormalities and genetic syndromes, malformation syndromes, non-genetic disorders that mimic genetic forms of the disease like amyloidosis and infiltrative diseases. In about 25-30% of HCM etiology is still unknown.

In adults, HCM is defined by a wall thickness ≥ 15 mm in one or more LV myocardial segments as measured by any imaging technique. Transthoracic 2D and tissue doppler echocardiography presents tool for morphologic and hemodynamic evaluation (with additional strain analysis, contrast and transesophageal echocardiography), and cardiac MRI for assessment of cardiac morphology and myocardial tissue characteristics. Routine laboratory tests and specific testing aids the detection of HCM etiology. The majority of HCM cases are inherited autosomal dominant genetic trait with a 50% risk of transmission to offspring. Some cases are explained by de novo mutations, sporadic cases can arise because of incomplete penetrance in a parent and by autosomal recessive inheritance. Regular genetic analysis should include the most commonly implicated sarcomere protein genes, and pedigree analysis should be provided. Annual incidence of cardiovascular death is 1-2% caused by sudden cardiac death, heart failure or thromboembolism.

HCM etiology detection is necessary because of treatment possibilities (like Anderson-Fabry disease or ATTR amyloidosis), detection of secondary causes of disease, the need for family screening and differentiating pathogenic from non-pathogenic mutations. Routine clinical practice is challenging, left ventricular hypertrophy and HCM can be presented in different forms and different stages.

RECEIVED:
February 28, 2019

ACCEPTED:
March 24, 2019



LITERATURE

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