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Pediatric hypertrophic cardiomyopathy: current diagnostic strategies, genetic insights, and challenges in risk stratification (a literature review)

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Abstract

Background. Pediatric hypertrophic cardiomyopathy (HCM) is a major cause of sudden cardiac death in children and represents a clinically and genetically heterogeneous condition distinct from adult disease. Although advances in adult HCM have informed management strategies, pediatric-specific evidence remains comparatively limited, particularly regarding imaging-based risk assessment and validation of pediatric risk prediction models.

Aim: To assess current diagnostic approaches, genetic insights, and current challenges in risk stratification in pediatric HCM.

Methods. A structured narrative review of studies published from 1997 through early 2025 was conducted using major biomedical databases and guideline references. Priority was given to pediatric cohort studies, population-based registries, contemporary clinical guidelines, and genetic investigations addressing epidemiology, etiologic classification, diagnostic evaluation, and risk prediction.

Key Content and Findings. Recent evidence confirms the predominant role of sarcomeric gene variants beyond infancy, while underscoring the importance of syndromic and metabolic etiologies in early childhood. Advances in electrocardiographic risk markers, multimodal imaging including cardiac magnetic resonance imaging assessment of myocardial fibrosis and next generation genetic testing have enhanced diagnostic precision and family screening. However, phenotypic variability, age-dependent penetrance, and limitations in external validation of pediatric risk prediction tools, including HCM Risk-Kids, continue to challenge individualized management.

Conclusion. Pediatric HCM remains a prognostically complex and biologically diverse disease. Refinement of pediatric-specific risk stratification models, improved integration of imaging and genetic data, and international collaborative research are essential to optimize early diagnosis and personalized care in affected children.

Keywords: pediatric hypertrophic cardiomyopathy; risk stratification; cardiac magnetic resonance imaging; genetic testing; sudden cardiac death.

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Детская гипертрофическая кардиомиопатия: современные диагностические стратегии, генетические аспекты и проблемы стратификации риска (обзор литературы)

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Аннотация

Введение. Детская гипертрофическая кардиомиопатия (ГКМ) является основной причиной внезапной сердечной смерти у детей и представляет собой клинически и генетически гетерогенное состояние, отличное от заболевания у взрослых. Несмотря на то, что достижения в изучении ГКМ у взрослых повлияли на стратегии лечения, данные, специфичные для детей, остаются относительно ограниченными, особенно в отношении оценки риска на основе визуализации и валидации прогностических моделей для детей.

Цель: Оценить современные диагностические подходы, генетические аспекты и текущие проблемы в стратификации риска при детской ГКМ.

Методы. Проведен структурированный нарративный обзор исследований, опубликованных с 1997 года по начало 2025 года, с использованием основных биомедицинских баз данных и ссылок на руководства. Приоритет отдавался педиатрическим когортным исследованиям, популяционным регистрам, современным клиническим руководствам и генетическим исследованиям, касающимся эпидемиологии, этиологической классификации, диагностической оценки и прогнозирования риска.

Основное содержание и результаты. Недавние данные подтверждают преобладающую роль вариантов генов саркомеров после младенческого возраста, подчеркивая при этом важность синдромальных и метаболических этиологий в раннем детстве. Достижения в области электрокардиографических маркеров риска, мультимодальной визуализации, включая оценку миокардиального фиброза с помощью магнитно-резонансной томографии сердца, и генетического тестирования нового поколения повысили точность диагностики и скрининга семей. Однако фенотипическая вариабельность, возрастная пенетрантность и ограничения внешней валидации инструментов прогнозирования риска в педиатрии, включая HCM Risk-Kids, продолжают создавать трудности для индивидуализированного лечения.

Заключение. Детская ГКМ остается заболеванием со сложным прогнозом и биологическим разнообразием. Усовершенствование прогностических моделей, специфичных для детей, улучшение интеграции данных визуализации и генетических данных, а также международные совместные исследования необходимы для оптимизации ранней диагностики и персонализированного ухода за пораженными детьми.

Ключевые слова: детская гипертрофическая кардиомиопатия; стратификация риска; магнитно-резонансная томография сердца; генетическое тестирование; внезапная сердечная смерть.

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Introduction

Pediatric hypertrophic cardiomyopathy (HCM) is the second most common cardiomyopathy in children and an important cause of sudden cardiac death (SCD) [1, 2]. It is defined by left ventricular hypertrophy (LVH) occurring in the absence of abnormal loading conditions and is frequently associated with myocardial disarray and fibrosis [3].

Although HCM has been extensively investigated in adults, pediatric disease remains comparatively underrepresented in clinical trials and registries. Importantly, pediatric HCM differs from adult forms in its etiologic spectrum, with a higher proportion of syndromic and metabolic causes, as well as age-dependent phenotypic expression and variable penetrance [4]. These differences complicate early diagnosis, risk stratification, and long-term management.

This narrative review summarizes current advances in the epidemiology, clinical presentation, diagnostic strategies, and genetic determinants of pediatric HCM, with emphasis on pediatric-specific considerations and implications for individualized care.

Methods

This narrative review synthesizes contemporary evidence on pediatric HCM. A structured literature search was conducted using PubMed/MEDLINE and Embase databases, supplemented by manual screening of reference lists from relevant articles and international clinical guidelines. Studies published between 1997 and early 2025 were considered.

Priority was given to peer-reviewed original research, large pediatric cohort studies, population-based registries, contemporary international guidelines, and

genetic investigations with direct relevance to pediatric populations. Studies focusing exclusively on adult cohorts without pediatric-specific analysis were excluded unless they provided mechanistic or methodological insights applicable to children.

No formal systematic review protocol or meta-analytic approach was applied. Instead, evidence was selected based on methodological rigor, clinical relevance, and contribution to advancing understanding of epidemiology, etiologic heterogeneity, diagnostic strategies, imaging-based risk assessment, and genetic testing in pediatric HCM. Findings were synthesized thematically to provide a critical overview of current knowledge and remaining challenges in pediatric risk stratification.

Epidemiology of pediatric hypertrophic cardiomyopathy

HCM accounts for approximately 40% of cardiomyopathies in children [5]. In contrast to the well-established adult prevalence of approximately 1:500 [6], the true prevalence of pediatric HCM remains uncertain. Population-based studies from North America, Australia, and Europe estimate an annual incidence ranging from 0.24 to 0.47 per 100,000 children [7–9].

Data from the Pediatric Cardiomyopathy Registry (PCMR) reported an incidence of 0.47 per 100,000 children in selected regions of the United States, with incidence peaks during infancy and adolescence [7]. Reported differences across studies likely reflect variations in genetic background, diagnostic criteria, case ascertainment, and study periods. Moreover, most available epidemiological data originate from high-income countries, while population-based data from developing regions remain scarce. This highlights the need for broader international registries and standardized diagnostic criteria to better define the global burden of pediatric HCM.

Table 1 summarizes data from major population-based pediatric cardiomyopathy registries, indicating the total number of cardiomyopathy cases and the proportion diagnosed with HCM.

Table 1. Characteristics of epidemiological studies on pediatric pediatric hypertrophic cardiomyopathy (data extracted from references) [7–9]

Таблица 1. Характеристики эпидемиологических исследований детской гипертрофической кардиомиопатии (данные, извлеченные из литературных источников) [7–9]

Region/Study	Study period	Total pediatric cardiomyopathy cases	No. of children with HCM	Percentage	Incidence (per 100,000 children)
Study in Finland	1980–1991	118	44	37%	0.24
New England & Southwest (PCMR)	1996–1999	467	196	42%	0.47
National study in Australia	1987–1996	314	80	25%	0.32

Note: The column “Total number of pediatric cardiomyopathy patients included in the study” refers to the overall number of pediatric cardiomyopathy cases enrolled in each population-based registry or study, from which HCM cases were identified.

Despite advances in diagnosis, mortality remains significant, and epidemiological data from low- and middle-income countries are still limited, underscoring the need for broader international registries.

Recent large cohort analyses published in 2025 have further highlighted the heterogeneity of clinical trajectories in pediatric HCM, demonstrating that outcomes vary significantly according to etiologic subgroup and age at presentation, reinforcing the importance of etiologic classification in prognostic assessment [10].

Etiologic Spectrum of pediatric hypertrophic cardiomyopathy

Pediatric HCM encompasses a heterogeneous group of disorders with variable clinical expression. In contrast to adult HCM, pediatric cases show a broader etiologic spectrum, including both sarcomeric and non-sarcomeric causes (Figure 1) [11, 12].

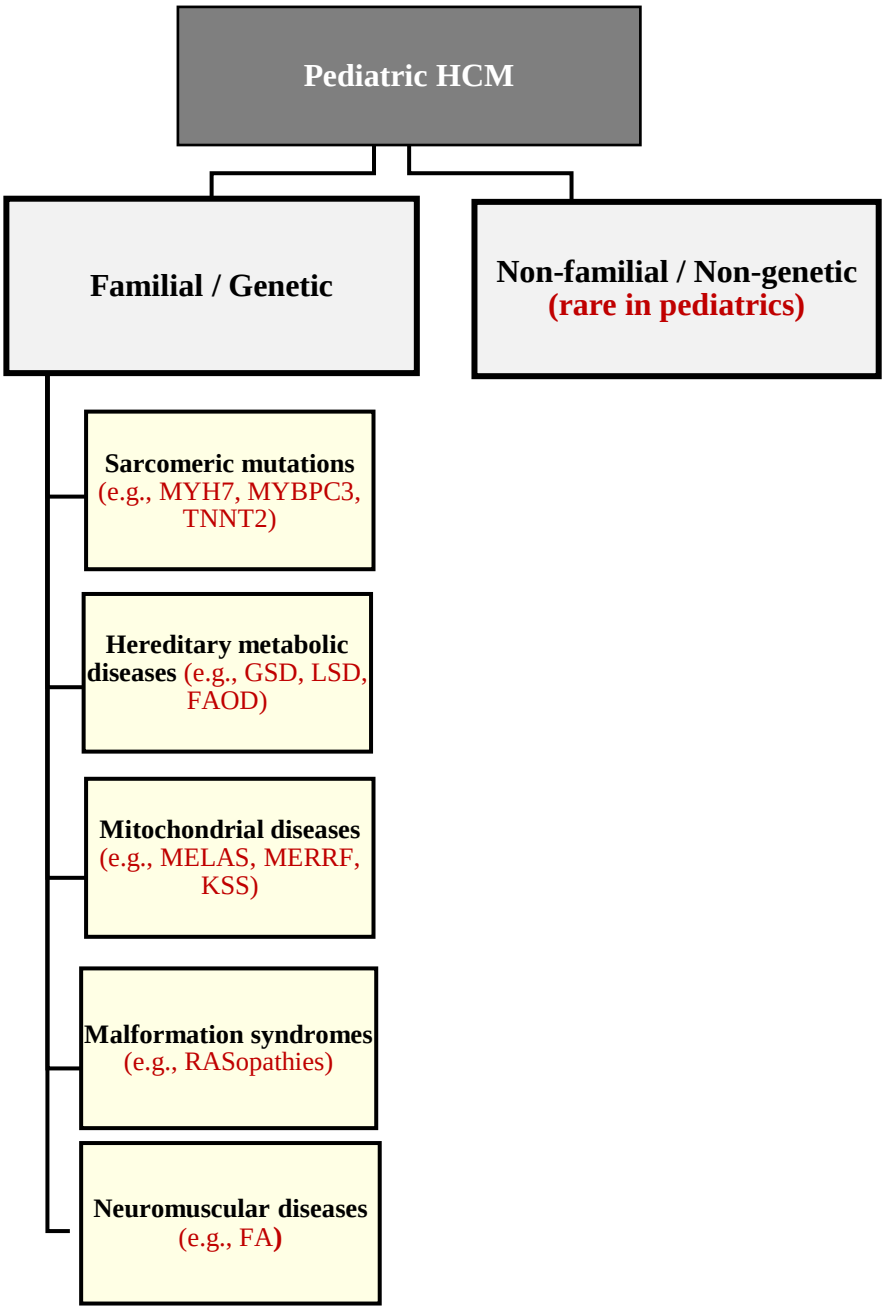

Fig. 1. Etiological classification of pediatric hypertrophic cardiomyopathy

Рис. 1. Этиологическая классификация детской гипертрофической кардиомиопатии

Sarcomeric gene variants remain the most frequent cause beyond infancy and are typically inherited in an autosomal dominant manner with age-dependent penetrance.

Age-dependent penetrance represents a key feature of sarcomeric Pediatric HCM. Genotype-positive children may remain phenotypically negative for prolonged periods, underscoring the importance of longitudinal clinical and imaging surveillance in at-risk individuals [11, 13].

The most commonly implicated genes include MYH7, MYBPC3, TNNT2, and TNNI3 [14, 15]. Recent registry data have demonstrated that sarcomeric disease in children may be more common than previously recognized and can be associated with significant morbidity [16].

Non-sarcomeric causes are particularly relevant in infancy and early childhood and include metabolic disorders (e.g., glycogen storage diseases), mitochondrial diseases, RASopathies such as Noonan syndrome (NS), and neuromuscular conditions [1, 17]. These etiologies often present earlier and may be associated with systemic manifestations and less favorable outcomes.

Accurate etiologic classification is essential for prognosis, genetic counseling, and family screening.

Clinical Presentation and Outcomes

The clinical presentation of pediatric HCM is highly variable, ranging from asymptomatic cases detected during family screening to progressive heart failure (HF), arrhythmias, and SCD [3, 4]. Many children remain asymptomatic for prolonged periods, while symptomatic patients may present with exertional dyspnea, chest pain, syncope, palpitations, or exercise intolerance.

In infancy, pediatric HCM often manifests as congestive heart failure (CHF) with feeding difficulties, poor weight gain, and respiratory distress, and this age group carries the highest mortality risk [18, 19]. In older children and adolescents, symptoms are more frequently related to left ventricular outflow tract (LVOT) obstruction, diastolic dysfunction, or arrhythmias.

In cases of significant LVOT obstruction, particularly when symptoms persist despite medical therapy, surgical management—most notably septal myectomy—represents an important therapeutic option in selected pediatric patients. Recent studies have highlighted the effectiveness of extended septal myectomy in improving LVOT gradients and clinical outcomes, including in complex and syndromic forms such as NS [20, 21].

Syncope may reflect hemodynamic compromise or ventricular arrhythmias and warrants careful risk assessment [4].

Although overall survival in children with HCM is generally better than in other forms of pediatric cardiomyopathy, disease progression and the risk of SCD remain major concerns, requiring regular follow-up and individualized management [1, 22].

Recent efforts have focused on the development of pediatric-specific risk prediction tools. The HCM Risk-Kids model represents a major step toward pediatric-specific risk stratification, integrating clinically relevant variables including maximal LV wall thickness, left atrial diameter, unexplained syncope, non-sustained ventricular tachycardia, and family history of SCD [23]. In the original derivation cohort, the model demonstrated moderate discriminative performance, supporting its utility in estimating individualized 5-year SCD risk.

However, subsequent external analyses have highlighted variability in model calibration across different populations, suggesting that predictive accuracy may depend on cohort characteristics and baseline risk distribution [24, 25]. Importantly, the derivation dataset was largely composed of patients from tertiary referral centers, potentially enriching the cohort with more severe phenotypes and limiting generalizability to lower-risk or community-based settings [23, 26]. In addition, patients with syndromic or metabolic forms of HCM were underrepresented, raising concerns regarding applicability across the full etiologic spectrum of pediatric HCM.

These considerations underscore that, while HCM Risk-Kids represents a significant advance, its performance must be interpreted within clinical context, and ongoing validation and refinement in diverse pediatric populations remain essential.

1. Diagnostic Evaluation

The diagnosis of pediatric HCM relies on a comprehensive clinical and multimodal imaging assessment. Evaluation may be initiated by symptoms, detection of a heart murmur, abnormal ECG findings, or systematic family screening in relatives of affected individuals [24, 27].

A 12-lead ECG is frequently abnormal and may demonstrate LVH, repolarization changes, pathological Q waves, or signs of left atrial enlargement (LAE). Although not specific, ECG abnormalities may precede overt hypertrophy and contribute to risk assessment in combination with clinical and imaging parameters [25, 26].

Recent data published in 2025 have further characterized ECG patterns in RASopathy-associated pediatric HCM, identifying specific electrocardiographic markers associated with adverse cardiovascular outcomes [28]. In particular, pathological Q waves, marked repolarization abnormalities with deep negative T waves, QRS fragmentation, and abnormal frontal QRS axis deviation were independently associated with major adverse cardiac events in this subgroup [28]. These findings suggest that ECG abnormalities may provide incremental prognostic information beyond their traditional diagnostic role, particularly in syndromic pediatric populations.

Transthoracic echocardiography remains the primary diagnostic modality. It allows precise measurement of ventricular wall thickness, assessment of LVOT obstruction, and evaluation of systolic and diastolic function [13, 29]. In children, wall thickness must be interpreted using body-size–adjusted Z-scores to avoid over- or underestimation of disease severity. Current guidelines recommend periodic screening in at-risk children, with follow-up intervals adapted according to age, phenotype evolution, and family history [13, 30].

Cardiac magnetic resonance imaging (MRI) provides complementary structural and tissue characterization, particularly in cases with suboptimal echocardiographic windows or suspected atypical patterns of hypertrophy. Late gadolinium

enhancement (LGE) enables detection of focal myocardial fibrosis and has emerged as a potential marker of disease severity [31, 32].

Beyond the mere identification of fibrosis, pediatric data suggest that LGE is associated with ventricular arrhythmias and indicators of disease progression [32]. Longitudinal observations further indicate that increasing LGE burden over time may reflect progressive myocardial remodeling and a more advanced phenotype [31, 32]. Although quantitative risk thresholds remain less clearly established in pediatric populations than in adults, greater LGE extent appears to correlate with more severe clinical presentation and may provide incremental prognostic value when integrated with clinical, echocardiographic, and rhythm monitoring findings [31].

Genetic Testing Strategies

Genetic testing is a cornerstone of the diagnostic evaluation of pediatric HCM. Identification of a pathogenic variant confirms the diagnosis in uncertain cases, facilitates cascade screening of relatives, and contributes to risk stratification and genetic counseling [33, 34].

Most pathogenic variants involve sarcomeric genes, particularly MYH7 and MYBPC3, although non-sarcomeric and syndromic causes are more frequent in early childhood [14, 35]. The main genes associated with pediatric HCM are summarized in Table 2. These include core sarcomeric genes responsible for the majority of familial cases, as well as genes implicated in metabolic, syndromic, and neuromuscular forms of the disease.

Table 2. Major genes associated with pediatric hypertrophic cardiomyopathy

Таблица 2. Основные гены, ассоциированные с детской гипертрофической кардиомиопатией

Category	**Representative genes**	**Clinical relevance**
Core sarcomeric genes	MYH7, MYBPC3, TNNT2, TNNI3	Most common causes beyond infancy; autosomal dominant inheritance
Additional sarcomeric / structural genes	TPM1, ACTC1, MYL2, MYL3, TNNC1	Less frequent familial forms

Metabolic / storage disorders	GAA, LAMP2, GLA, PRKAG2	Often early onset; may present with systemic manifestations
RASopathies / syndromic forms	PTPN11, RAF1, SOS1, KRAS	Associated with Noonan spectrum disorders
Mitochondrial / neuromuscular disorders	FXN, DES	Multisystem involvement; variable prognosis

Genetic testing strategies have evolved from Sanger sequencing of selected genes to next-generation sequencing (NGS)–based multigene panels, which are now considered first-line diagnostic tools due to their broader coverage and cost-effectiveness [36, 37].

The diagnostic yield of multigene panels in pediatric HCM ranges from approximately 40% to 60%, depending on age at presentation and family history. Higher yields are typically observed in familial and early-onset cases, whereas genotype-negative patients may reflect undiscovered variants, complex inheritance patterns, or limitations of current testing strategies [35, 38].

Whole exome sequencing (WES) and whole genome sequencing (WGS) may increase diagnostic yield in selected cases, particularly in patients who remain genotype-negative after targeted testing. However, these approaches generate a higher number of variants of uncertain significance (VUS) and require careful interpretation within clinical context [39, 41].

Overall, genetic testing plays a critical role in etiologic classification, family screening, and personalized management, although genotype–phenotype correlations remain incompletely understood.

Emerging Epigenetic Mechanisms

Beyond pathogenic gene variants, epigenetic mechanisms such as DNA methylation and non-coding RNAs have been increasingly implicated in cardiac hypertrophy and myocardial remodeling. Experimental and translational studies suggest that epigenetic modifications may influence gene expression, disease progression, and phenotypic variability in HCM [42, 43]. However, pediatric-specific

evidence remains limited, and the clinical utility of epigenetic biomarkers in pediatric HCM is not yet established.

Conclusion

Pediatric HCM remains a prognostically complex and biologically heterogeneous disease, characterized by substantial etiologic diversity and age-dependent phenotypic expression. Although advances in electrocardiographic markers, cardiac MRI-based fibrosis assessment, and next-generation genetic testing have improved diagnostic precision and family screening, important uncertainties persist regarding individualized risk prediction in children.

Current pediatric risk stratification models, including HCM Risk-Kids, represent significant progress but require continued validation across diverse populations and etiologic subgroups. The integration of clinical, imaging, and genetic data remains essential to refine prognostic assessment and guide personalized management strategies.

Future pediatric-focused research and international collaborative registries are crucial to better define genotype-phenotype correlations, improve external validation of risk prediction tools, and optimize long-term outcomes in affected children.

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