RESEARCH ARTICLE

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Pediatric Left Ventricular Non-compaction Cardiomyopathy: Radiological and Echocardiographic Imaging Findings

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Abstract

Objectives: Non-compaction cardiomyopathy is a rare form characterized by the ventricular myocardium comprising an outer layer of normally compacted myocardium and an inner layer of non-compacted myocardium. The diagnosis primarily relies on imaging modalities, including echocardiography (ECHO) and cardiac magnetic resonance imaging. The aim of our study was to evaluate ECHO and cardiac magnetic resonance imaging findings in a group of children with isolated left ventricular non-compaction.

Methods: Between September 2022 and January 2025, pediatric patients under 18 years of age, who exhibited ECHO findings suggestive of left ventricle non-compaction, were retrospectively enrolled. Contrast-enhanced cardiac magnetic resonance imaging, including late gadolinium enhancement as well as two-chamber, four-chamber, and short-axis views, was performed to assess ventricular size, wall characteristics, and function in patients with suspected myocardial non-compaction based on ECHO findings.

Results: A total of 35 patients, with a median age of 14 years, were recruited. In the 12 patients where both ECHO and cardiac magnetic resonance imaging (cMRI) findings were concordant, leading to a definitive diagnosis on ECHO, the median non-compacted-to-compacted (NC/C) myocardial layer ratio in diastole was 2.6, while on cMRI, according to Petersen's criteria, the median NC/C ratio in end-diastole was 1.6.

Conclusion: ECHO is a reliable and non-invasive modality for monitoring left ventricular systolic function and wall characteristics. However, cardiac magnetic resonance imaging is recommended for a more precise assessment of left ventricular remodeling, right ventricular size and function, and the detection of myocardial fibrosis.

Keywords: Cardiomyopathy, left ventricular non-compaction, echocardiography, cardiovascular magnetic resonance imaging, children

Introduction

Non-compaction cardiomyopathy is a rare form of cardiomyopathy characterized by the ventricular myocardium comprising an outer layer of normally compacted myocardium and an inner layer of non-compacted myocardium.[1,2] The left ventricular myocardium consists of two distinct layers: a compact layer and a non-compact layer.[3] Non-compaction is traditionally associated with the left ventricle but may also involve the right ventricle as part of a biventricular presentation.[4,5] The American Heart Association (AHA) classifies left ventricular non-compaction (LVNC) as a genetic cardiomyopathy,[6] whereas the European Society of Cardiology categorizes it as a condition associated with other cardiomyophatie phenotypes (unclassified) and left ventricule hypertrabeculation.[7]

LVNC may present as an isolated condition or associated with congenital heart diseases, genetic syndromes, or neuromuscular disorders. This observation supports the notion that LVNC is not a distinct cardiomyopathy but rather a morphological manifestation of various underlying diseases.[1]

LVNC is often asymptomatic, but it can manifest with heart failure, arrhythmias, and thromboembolic events. Electrocardiography (ECG) abnormalities are more common in pediatric patients; however, they are non-specific.[2]

Although echocardiography (ECHO) and cardiac magnetic resonance imaging (cMRI) are widely utilized, the gold standard diagnostic criteria remain undefined due to the reversibility of the hypertrabecular structure and the variability in systolic and diastolic function findings among patients.[3] The diagnosis of LVNC is based on morphological criteria. ECHO is the primary diagnostic modality, and also essential for patient follow-up. cMRI with late gadolinium enhancement (LGE) is a complementary imaging tool that aids in confirming the diagnosis and provides prognostic insights.[8]

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cMRI has gained increasing popularity in recent years as a diagnostic imaging modality due to its high-resolution anatomical detail. It allows for a comprehensive assessment of systolic and diastolic function, detection of myocardial fibrosis through LGE, and evaluation of ventricular wall structure and motion. Numerous studies have examined cMRI's diagnostic utility and criteria in identifying LVNC in adults.[2,5,9] These investigations consistently demonstrate the superiority of cMRI over ECHO, particularly in evaluating myocardial compaction in regions that are challenging to visualize with ECHO, such as the left ventricular apex and lateral wall.[9] In routine clinical practice, the most widely accepted cMRI-based diagnostic criterion for LVNC in adults is a non-compacted to compacted (NC/C) myocardium ratio of ≥2.3 during end-diastole, as established by Petersen et al.[10] in a cohort of patients aged 14 to 46 years. In contrast, limited research has explored the diagnostic role of cMRI in pediatric populations, and standardized diagnostic criteria for children have yet to be established.[3,8] The objective of the present study is to assess cMRI findings in pediatric patients with LVNC, to compare these with ECHO findings, and to evaluate the applicability of adult-based diagnostic criteria in a pediatric context, with the aim of proposing appropriate criteria for use in children.

Methods

Patients and Data Collection

From September 2022 to January 2025, pediatric patients with suspicious ECHO features for LVNC will be retrospectively screened and included in the study. Poor imaging quality, as well as congenital heart disease, other types of cardiomyopathies, or neuromuscular disorders in patients, were exclusion criteria for the study. cMRI images were evaluated for the presence of LVNC.

The cMRI, ECHO, and electrophysiological findings of patients with LVNC were recorded, as were the patients' demographic characteristics, clinical symptoms, and family history of cardiomyopathy and sudden cardiac death (SCD).

The study was approved by University of Sciences Türkiye, Ankara Etlik City Hospital's Clinical Research Ethics Committee (approval number: 2024-765, date: 02.12.2024).

Echocardiographic Imaging

ECHO imaging was performed using two-dimensional, Doppler, and M-mode ECHO. Parasternal long-axis and short-axis views, as well as apical two-chamber, three-chamber, and four-chamber views, were acquired. The criteria for inclusion in the study were an age of less than 18 years at the time of diagnosis and ECHO evidence of isolated LVNC, defined by ECHO measurements evaluated according to Pignatelli's criteria: (1) the presence of multiple ECHO trabeculations, (2) multiple deep intertrabecular recesses communicating with the ventricular cavity, as demonstrated by color Doppler imaging, with recesses observed in the apical or mid-ventricular regions, and (3) a two-layered myocardial structure with >1.4 at end diastole phase.[8] NC/C ratio was measured in the parasternal short-axis view during the end-diastolic phase at three locations within the posterior and posterolateral regions of the left ventricle (Figure 1).

Cardiac Magnetic Resonance Imaging

cMRI was conducted using a "1.5-T scanner (Ingenia Evolution, Philips Medical Systems, Best, The Netherlands)", equipped with a dedicated cardiac phased-array coil and ECG gating. Steady-state free precession CINE sequences were obtained in the short-axis, four-chamber, and two-chamber planes. LGE imaging was performed in both the short-axis and the 2- and 4-chamber planes, commencing 10 minutes after administering 0.1 mmol/kg of gadobutrol. Image analysis was performed using the Philips IntelliSpace Portal cardiac imaging software. cMRI was evaluated by two radiologists (Ş.Y, H.U.) with at least 5 years of experience in this field.

In the cMRI examination, axial, sagittal, and coronal white blood sequences were acquired in orthogonal planes to assess anatomical details. CINE imaging was performed in the two-chamber, four-chamber, and short-axis views to evaluate left ventricular ejection fraction (EF), end-diastolic volume (EDV), end-systolic volume (ESV), and ventricular wall structure. The endocardial and epicardial borders of the left ventricle were delineated in these phases to determine EDV and ESV for both ventricles, enabling the calculation of EF and stroke volume (SV). The assessment of NC/C myocardial layers was performed by measuring their thickness perpendicular to the compacted myocardium in the EDV using long-axis and short-axis views. Measurements excluded the 17th segment, in accordance with the AHA model. The highest NC/C ratio was recorded (Figure 2).

In this study, the thickness of the most hypertrophied myocardial segment in the mid-section of the interventricular septum was measured in both short-axis and long-axis images of patients with suspected LVNC. This assessment aimed to determine whether septal thickness could be a diagnostic parameter in suspected cases. EDV and ESV were indexed to

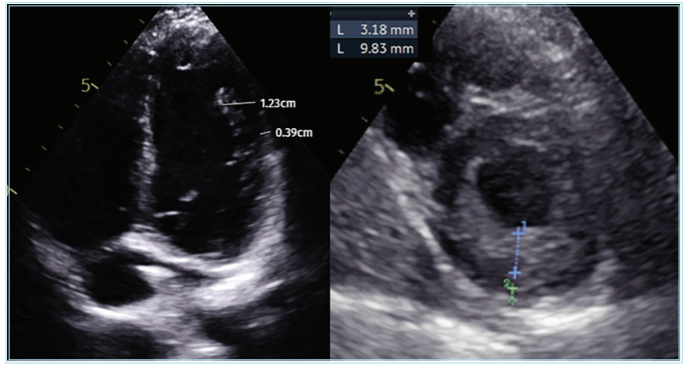

Figure 1. In echocardiographic images of the left ventricle, an increased non-compacted-to-compacted ratio was observed in measurements obtained during the end-diastolic phase, in the right long-axis and left short-axis views

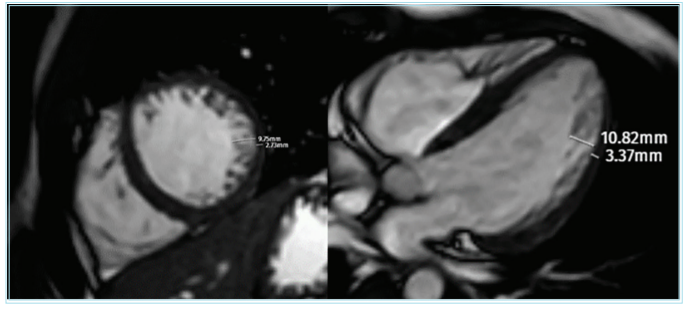

Figure 2. In cardiac magnetic resonance imaging images with white blood CINE imaging, an increased non-compacted-to-compacted ratio was observed in the left ventricular free wall, in the (right) short-axis and (left) long-axis views

the patient's body surface area to assess morphological abnormalities. They were compared with recently published normative values for children and adolescents, obtained using the same methodology.[11] In addition, all studies were visually evaluated for the presence of myocardial LGE.

Statistical Analysis

The distribution of all continuous variables was assessed using the Shapiro-Wilk test. Normally distributed variables are presented as mean±standard deviation, whereas non-normally distributed parameters are presented as median (interquartile range). The Pearson correlation coefficient was used to compare the LV non-compaction/compaction ratio between imaging modalities and to compare the cMRI non-compaction/compaction ratio with other findings. Statistical Package for the Social Sciences 26.0 (IBM, Chicago, IL, USA) was used to analyze all statistical data. A p value <0.05 was considered statistically significant.

Results

Clinical Findings

We reviewed the cMRI images of 32 patients, with ECHO features that were suspicious of LVNC. Due to technical problems and motion artifacts, 5 (16%) non-diagnostic examinations were excluded from the evaluation. In addition, 2 (6%) patients with congenital heart disease were excluded from the study. Thirteen (41%) patients did not meet the criteria for LVNC on cMRI. Demographic data for the 12 (38%) patients diagnosed with LVNC, confirmed through ECHO and CMR, were analyzed (Table 1).

Table 1. LVNC diagnosed patients' mean values of dermographic data

Baseline characteristics	n=12
Sex (boys/girls)	(12/6)
Age (years) median, IQR	15 (12.5-16)
Height (cm) mean±SD	161.5±19.9
Weight (kg) mean±SD	53.5±16.9
BMI (kg/m^2) mean±SD	20.1±3.3
BSA (m^2) mean±SD	1.53±0.34

LVNC: Left ventricular non-compaction, IQR: Interquartile range, SD: Standard deviation, BMI: Body mass index, BSA: Body surface area

The age range of patients with ECHO features suggestive of LVNC is 6-18 years, with a mean age of 14. Twenty patients (63%) were male, and 12 (37%) were female. The primary reasons for referral included clinical symptoms such as dyspnea (n=1), cardiac arrhythmias (n=4), chest pain (n=10), syncope (n=3), dizziness (n=3), heart murmur (n=2), as well as routine sports license evaluations (n=11) and a family history of cardiomyopathy or SCD. A total of 5 (16%) patients had a family history of early SCD of unknown etiology among first-degree relatives, while 3 (9%) patients had a diagnosis of dilated cardiomyopathy.

Electrocardiography and Ecocardiographic Findings

A total of 8 (25%) patients exhibited abnormal ECG findings, including incomplete right bundle branch block (n=2, 25%), atrioventricular block with sinus tachycardia (n=2, 25%), bradycardia (n=2, 25%), and ectopic beats (n=2, 25%). Among these, 1 (13%) patient with bradycardia had only ECHO abnormalities. All other patients with ECG abnormalities demonstrated positive findings for LVNC on both ECHO and cMRI.

Our study evaluated all 35 patients, who underwent cardiac cMRI based on ECHO findings. Among them, 10 patients had an NC/C ratio of 1-1.4 in the end-diastolic phase and were referred for cMRI due to high clinical suspicion. However, no pathological findings were detected on cMRI in these patients, and they were subsequently monitored. Additionally, among the 12 patients who met the diagnostic criteria on both cMRI and ECHO, mitral valve prolapse was identified in 3 patients, while 1 patient exhibited concomitant tricuspid and pulmonary insufficiency. No significant additional ECHO abnormalities were detected in the remaining patients.

Cardiac Magnetic Resonance Imaging Findings

In 13 patients with NC/C values of 1.4 or higher on ECHO (mean 1.6), while cMRI findings were negative the mean NC/C ratio measured on MRI was also 1.6. In contrast, 12 patients demonstrated positive findings for LVNC on both ECHO (mean NC/C 1.8) and cMRI (mean NC/C 2.6) (Table 2). LGE was not detected in any patient on cMRI.

Table 2. cMRI and echocardiographic findings in patients diagnosed with LVNC based on combined cMRI and echocardiographic evaluation

Patient no	Age	BMI	EDVI	ESVI	EF	IVS	ECHO NC/C	cMRI NC/C
1	15	22.4	62	21	67	7.5	3.1	3.6
2	12	15.4	20	19	68	6.1	1.5	2.5
3	16	21.6	63	26	63	7.7	1.7	2.3
4	16	22.2	90	36	60	5.3	1.4	2.9
5	15	20.9	80	36	59	5.6	1.6	2.6
6	14	26.7	77	38	62	4.6	2.6	2.7
7	18	17.3	64	20	69	6.7	1.8	2.7
8	15	22.1	55	23	58	4.5	1.8	2.7
9	16	19.1	61	29	62	6.1	1.7	2.4
10	8	20.8	43	16	76	6.6	1.4	2.3
11	8	16	45	18	64	6	1.5	2.3
12	15	16.6	104	34	55	5.6	1.9	3

cMRI: Cardiac magnetic resonance imaging, LVNC: Left ventricular non-compaction, BMI: Body mass index, EDVI: End diastolic volume index, ESVI: End systolic volume index, EF: Ejection fraction, IVS: Interventricular septum, ECHO: Echocardiography, NC/C: Non-compacted-to-compacted

Of the 12 patients who met the diagnostic criteria on both cMRI and ECHO, ECHO findings were similar in that mitral valve prolapse was observed in 3 patients. In contrast, 1 patient exhibited concomitant tricuspid and pulmonary insufficiency.

The NC/C ratios measured using ECHO correlate with those obtained through cMRI. This finding contrasts with a study conducted on adult patients, which reported a high degree of agreement between ECHO and cMRI measurements at end-diastole. Additionally, in the assessment of left heart function, EF, ESV, EDV, and SV, values indicated a mild reduction in left ventricular function in 3 of the 12 patients who met the definitive diagnostic criteria on cMRI (Table 3). In all examinations, a strong correlation was observed in NC/C ratios among patients who met the diagnostic criteria (Figure 3, Table 4).

Interventricular septum measurements were performed in LVNC patients to assess potential coexisting pathologies and their associations with other cardiomyopathies. Based on imaging studies conducted in these patients, no age-inappropriate abnormal septal thickness was detected.

Discussion

Although the classification of LVNC within the cardiomyopathy category remains a subject of debate among certain publishers and professional associations, ambiguities in its definition and diagnostic criteria do not dispute its clinical significance, as well as its disease progression, outcomes, and associated complications.[12]

LVNC can occur in both familial and sporadic forms. The familial recurrence rate in the pediatric population has been determined to be 50%, which is higher than that observed in the adult population.[13,14] In our study, genetic analysis was not conducted for family history; however, a family history of cardiac disease was identified in 25% of cases.

Table 3. cMRI and ECHO mean values for LVNC

ECHO and cMRI	Mean value
ECHO NC/C mean±SD	1.84±0.50
cMRI NC/C mean±SD	2.67±0.36
EDV (mL) mean, SD	105.5±29.06
ESV (mL) mean±SD	40.33±13.84
SV (mL) mean±SD	64.91±17.53
EDV/BSA (mL/m^2) mean±SD	67±12.68
ESV/BSA (mL/m^2) mean±SD	27.16±7.52
IVS (mm) mean±SD	6.02±1.01

cMRI: Cardiac magnetic resonance imaging, LVNC: Left ventricular non-compaction, ECHO: Echocardiography, SD: Standard deviation, EDV: End diastolic volume, ESV: End systolic volume, SV: Stroke volume, BSA: Body surface area, IVS: Interventricular septum

LVNC is more frequently observed in men, with reported prevalence ranging from 56% to 82%.[15,16] In the study conducted by Paszkowska et al.[17] on pediatric patients, the proportion of male patients diagnosed using cMRI was found to be 44%. In our study, this rate, with a male predominance, was determined to be 50% among a total of 12 patients who met the diagnostic criteria in both ECHO and cMRI examinations.

Abnormal ECG findings are frequently observed in pediatric patients with LVNC; however, they are often non-specific. Zuccarino et al.[18] reported that ECG changes may include ST-T abnormalities, bundle branch block, or Brugada-like ECG patterns. In the study conducted by Brescia et al.[19] on pediatric LVNC patients, arrhythmias were observed in 33.1% of 242 patients, with ventricular tachycardia identified in 17% of these cases. In our study, arrhythmias were observed in 7 out of 12 patients diagnosed with LVNC. Consistent with the literature, bundle branch block and sinus tachycardia were the most commonly detected arrhythmias. In the more advanced stages of LVNC, heart failure is observed in over 50% of patients.[5]

ECHO is the primary imaging method in cardiac evaluation in pediatric patients with its low cost, easy accessibility, and reproducibility.

In ECHO measurements studies, assessments have generally been conducted based on the Jenni criteria. However, these methods are employed as they are more suitable for pediatric patients.[7] Given that ECHO is a dynamic imaging modality with inherent subjectivity and operator dependence, cMRI correlation is particularly crucial in pediatric patients to ensure diagnostic accuracy and reliability.

In our study, in this context, 25 out of 35 patients met the diagnostic criteria based on ECHO evaluation, while 10 patients with high clinical suspicion underwent cMRI. Ultimately, 12 patients received a definitive

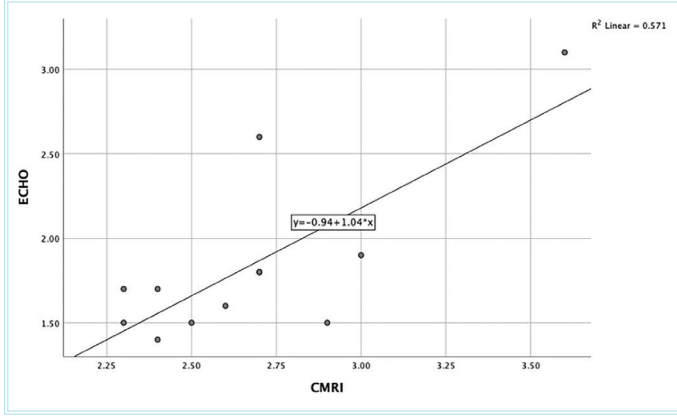

Figure 3. Non-compacted-to-compacted ratio cMRI and ECHO correlation analysis graph

ECHO: Echocardiography, cMRI: Cardiac magnetic resonance imaging

Table 4. Corelation between cMRI and ECHO

		ECHO NC/C	EDV	ESV	SV	EDV/BSA	ESV/BSA	EF	IVS
	Pearson r	**0.756***	0.20	0.13	0.23	0.217	0.064	0.08	0.05
cMRI NC/C	p	**0.004***	0.52	0.67	0.46	0.499	0.843	0.80	0.86
	n	**12**	12	12	12	12	12	12	12

*p<0.005.
cMRI: Cardiac magnetic resonance imaging, ECHO: Echocardiography, NC/C: Non-compacted-to-compacted, EDV: End diastolic volume, ESV: End systolic volume, SV: Stroke volume, BSA: Body surface area, EF: Ejection fraction, IVS: Interventricular septum

diagnosis based on the findings from both modalities. Notably, none of the 10 patients who could not be definitively diagnosed using ECHO met the diagnostic criteria on cMRI, which further demonstrates ECHO's diagnostic strength and reliability.

In the study conducted by Paszkowska et al.,[17] cMRI confirmed the diagnosis in 93% of children who exhibited LVNC features on ECHO. In our evaluation, this rate was measured as 48%.

Performing cMRI examination in all patients who meet the ECHO diagnostic criteria and are deemed highly suspicious for certain conditions is crucial for enhancing diagnostic accuracy. Additionally, it plays a significant role in patient follow-up, in treatment planning, and in identifying and monitoring comorbid conditions.

Our pediatric study compared ECHO images with those obtained via cMRI, both acquired at end-diastole to evaluate the NC/C ratio. The NC/C ratios measured using ECHO significantly correlated with those obtained through cMRI. This finding contrasts with a study conducted on adult patients, which reported a high degree of agreement between ECHO and cMRI measurements at end-diastole. This result is consistent with findings from a previous study conducted in pediatric patients.[1]

In the studies conducted by Grothoff et al.,[20] none of the cardiomyopathy patients exhibited LGE. In a cMRI study conducted on adult patients, which included a total of 47 individuals with LVNC, myocardial waves were detected in 40% of the patients, most frequently, in the mid-myocardial region.[21] In a study involving 25 pediatric patients, LGE was observed in 24% of cases, most commonly in the mid-myocardial region.[17] LGE was not detected in any of our patients. LGE represents fibrosis in these patients and can also indicate the possibility of suspected possible myocarditis. related. This rate is lower in pediatric patients than in others, possibly due to the relatively low development of fibrosis, which is attributed to the early onset of the disease.

In our study, the interventricular septum was measured at its thickest point in the midventricular region during the end-diastolic phase and assessed for its diagnostic contribution in patients with LVNC. Septal thickness was not found to exceed normal values in any of the diagnosed patients.

Study Limitations

This study is a single-center, retrospective analysis conducted over approximately three years at a large tertiary referral institution. As such, several limitations should be acknowledged. First, the sample size may limit the generalizability of the findings, as the characteristics of the study population may not be representative of broader or more diverse populations. Second, due to the retrospective design and the operator-dependent nature of ECHO assessments, a comprehensive analysis of diastolic function parameters and a more detailed evaluation of systolic function were not feasible. Finally, this study excluded patients under the age of six; consequently, assessment of noncompaction in this age group was not performed. This exclusion was due to the susceptibility of cardiac MRI sequences to motion artifacts, which are more prevalent in younger children.

Conclusion

cMRI is strongly recommended as a complementary imaging tool for assessing non-compaction, to accurately assess the extent of myocardial non-compaction and to evaluate ventricular size and systolic function reliably. To better understand the necessity and comparative value of ECHO and cMRI, further studies with larger patient cohorts are needed.

Ethics

Ethics Committee Approval: The study was approved by University of Health Sciences Türkiye, Ankara Etlik City Hospital's Clinical Research Ethics Committee (approval number: 2024-765, date: 02.12.2024).

Informed Consent: Because this was a retrospective study, informed consent was not required by the ethics committee.

Footnotes

Author Contributions

Surgical and Medical Practices: Ş.Y., Ö.K., Concept: Ş.Y., H.B., Design: Ş.Y., H.B., Ö.K., Data Collection or Processing: Ş.Y., Analysis or Interpretation: Ş.Y., H.B., Literature Search: Ş.Y., Ö.K., Writing: Ş.Y.

Conflict of Interest: No conflict of interest was declared by the authors.

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