Case Report

# Biventricular Noncompaction Cardiomyopathy Associated With Neuropathy in a Young Patient: A Case Report

*Mônica de Oliveira Gonçalo,[1] André Alexandre dos Santos Gomes,[1] Matheus Martins Monteiro,[2] Nicolas Babilônia Cavalcanti,[1] Fernando Almeida Bezerra,[1] Moisés Abtibol Machado,[1] Dênison Clark Corrêa de Miranda,[1] Kátia do Nascimento Couceiro,[1] João Marcos Bemfica Barbosa Ferreira[1]*

*Universidade do Estado do Amazonas,[1] Manaus, AM – Brazil*
*Beneficência Portuguesa de São Paulo,[2] São Paulo, SP – Brazil*

## Introduction

Ventricular hypertrabeculation, also known as ventricular noncompaction, is a myocardial phenotype characterized by excessive trabeculation and deep intertrabecular recesses caused by incomplete myocardial compaction during embryonic development. The earliest reported cases were associated with congenital heart defects, including ventricular outflow tract obstruction, complex cyanotic congenital heart disease, and coronary artery anomalies.[1,2]

The clinical presentation is highly heterogeneous and may include heart failure (HF), arrhythmias, thromboembolic events, and sudden cardiac death. In addition to ventricular structural abnormalities, ventricular hypertrabeculation may be associated with cardiac conduction disorders, atrial arrhythmias, and functional valvular regurgitation (FVR) secondary to cardiac chamber dilatation, all of which contribute to greater clinical complexity and a poorer prognosis.[3]

Furthermore, ventricular hypertrabeculation is a well-recognized feature of several neuromuscular disorders, suggesting a shared genetic basis and a syndromic phenotype in a subset of patients. Diagnosis may be challenging because of its marked phenotypic variability and overlap with other cardiomyopathies. Cardiac magnetic resonance (CMR) plays a particularly important role in confirming the diagnosis and providing detailed anatomical characterization.[4]

We report the case of a young patient with biventricular noncompaction associated with significant valvular heart disease, cardiac conduction disease requiring permanent pacemaker implantation, who is undergoing investigation for an underlying neuromuscular disorder. The patient is enrolled in a study approved by the Human Research Ethics Committee of the Universidade do Estado do Amazonas (approval no. 7,108,530; CAAE no. 55200322.5.3009.516).

## Keywords

Left Ventricular Hypertrophy; Mitral Valve Insufficiency; Case Reports

**Mailing Address: Matheus Martins •**
Beneficência Portuguesa de São Paulo. R. Maestro Cardim, 637. Postal code: 01323-900. Bela Vista, São Paulo, SP – Brazil
E-mail: matheussmartins.1996@gmail.com
Manuscript received May 25, 2026, revised manuscript June 2, 2026, accepted July 7, 2026
Editor responsible for the review: Marcelo Tavares

**DOI:** https://doi.org/10.36660/abcimg.20260078i

## Case report

A 21-year-old woman from the city of Tefé, state of Amazonas, Brazil, was referred to a specialized cardiomyopathy clinic with a diagnosis of biventricular noncompaction associated with severe mitral regurgitation (MR) and tricuspid regurgitation (TR) secondary to annular dilatation. Her medical history included permanent atrial fibrillation (AF) ($CHA_2DS_2$-VA score = 3; HAS-BLED score = 0), complete atrioventricular (AV) block with prior implantation of a dual-chamber permanent pacemaker, and a previous episode of compensated cardiogenic ascites. A neuromuscular disorder under investigation was the most relevant comorbidity.

Her clinical history revealed symptom onset during childhood, beginning with progressive ascites at 11 years of age. She was initially managed at a local health care facility but showed no satisfactory response to medical therapy. At that time, both cardiac and hepatic etiologies were considered, prompting referral to a tertiary referral center, where she remained hospitalized for approximately 3 months. Thereafter, outpatient follow-up was irregular, partly because it relied on telemedicine.

At 20 years of age, she experienced recurrent massive ascites refractory to diuretic therapy, with subsequent clinical improvement following evaluation at a private health care facility. At 21 years of age, she developed another episode of decompensation characterized by ascites and dyspnea, requiring hospitalization and referral to a tertiary care center. During this admission, the diagnosis of biventricular noncompaction cardiomyopathy was established.

On physical examination, the patient appeared underweight, with normal skin coloration, no cyanosis, and hypotension (blood pressure: 88/58 mmHg; heart rate: 71 bpm), without signs of peripheral congestion. Cardiac auscultation revealed a regular two-sound rhythm and a systolic murmur best heard at the pulmonary area. Pulmonary auscultation demonstrated normal vesicular breath sounds without adventitious findings, and no lower-extremity edema was present. Electrocardiography showed a junctional rhythm, extreme axis deviation, and left bundle branch block (Figure 1).

Transthoracic echocardiography demonstrated biatrial enlargement and mild eccentric left ventricular (LV) hypertrophy. Global LV systolic function was preserved (left ventricular ejection fraction [LVEF], 51%). Additional findings included myxomatous degeneration with prolapse

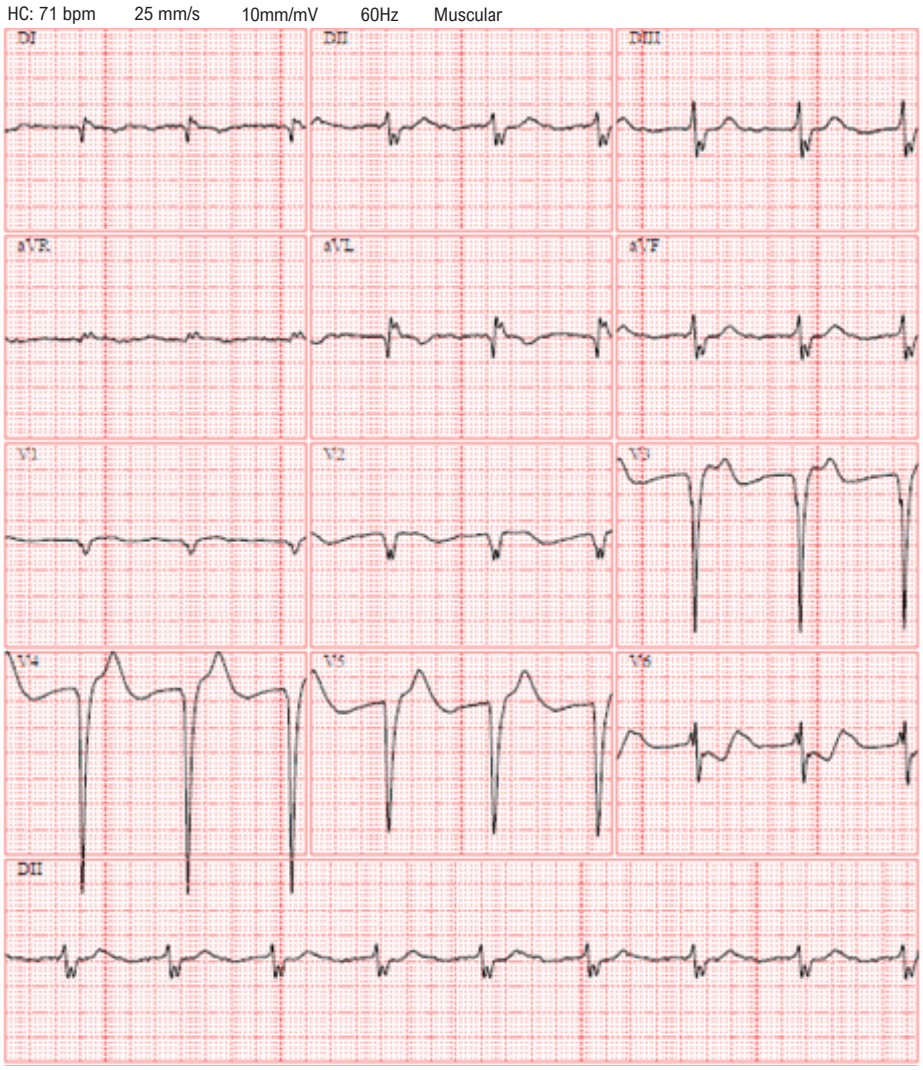


**Figure 1 –** *Electrocardiogram demonstrating a junctional rhythm, extreme axis deviation, and left bundle branch block. HR: heart rate.*

of the anterior mitral leaflet, resulting in severe MR, mild pulmonary hypertension, spontaneous echo contrast in the right atrium, and ventricular hypertrabeculation.

CMR demonstrated biventricular chamber enlargement (Figure 3), with indexed LV and right ventricular (RV) end-diastolic volumes of 110.33 mL/m$^2$ and 119.37 mL/m$^2$, respectively. Prominent LV myocardial trabeculation was observed, with a noncompacted-to-compacted myocardium ratio > 2.3, meeting the Petersen diagnostic criterion. LVEF was preserved (56%), whereas the RV exhibited systolic dysfunction (ejection fraction, 30%), accompanied by biatrial enlargement, functional MR, and a small pericardial effusion, without evidence of late gadolinium enhancement or intracardiac thrombi.

Neurological evaluation revealed a motor deficit syndrome involving all four limbs, muscle atrophy, and findings suggestive of a neuromuscular disorder with probable X-linked inheritance. Emery-Dreifuss muscular dystrophy (EDMD) was considered the leading diagnostic hypothesis. As part of the etiological investigation, G-banded karyotyping was performed and demonstrated a normal female chromosomal complement without detectable abnormalities.

At 25 years of age, the patient underwent dual-chamber permanent pacemaker implantation because of complete AV block. At her most recent outpatient follow-up, she reported occasional palpitations but denied chest pain, dyspnea, or peripheral edema. She also reported good adherence to guideline-directed medical therapy, consisting of enalapril, carvedilol, dapagliflozin, furosemide, and spironolactone, together with anticoagulation with rivaroxaban and fluid restriction.

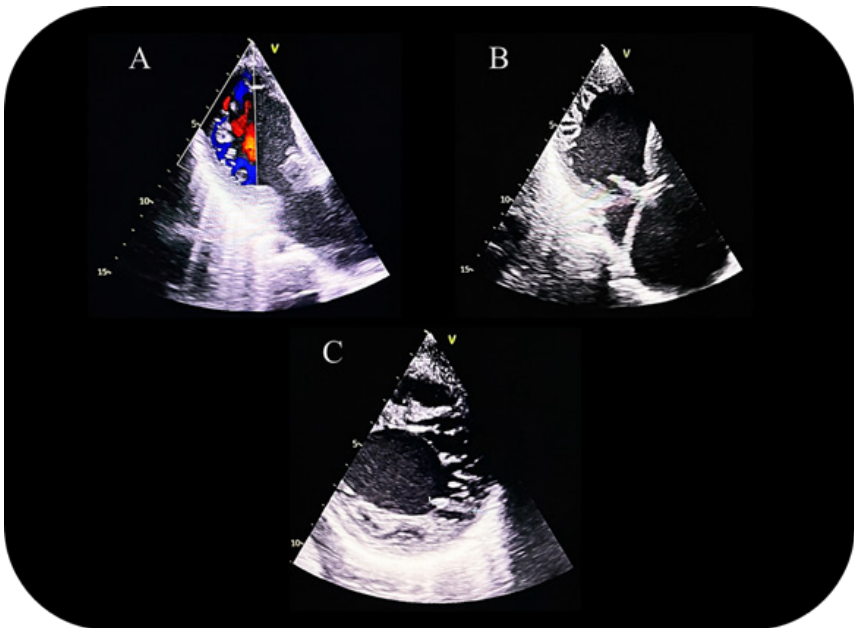


**Figure 2 –** *Transthoracic echocardiography demonstrating hypertrabeculation of the RV free wall. A) Apical view with color Doppler imaging. B) Two-dimensional apical view demonstrating prominent trabeculations. C) Parasternal short-axis view demonstrating hypertrabeculation of the RV free wall. RV: right ventricle.*

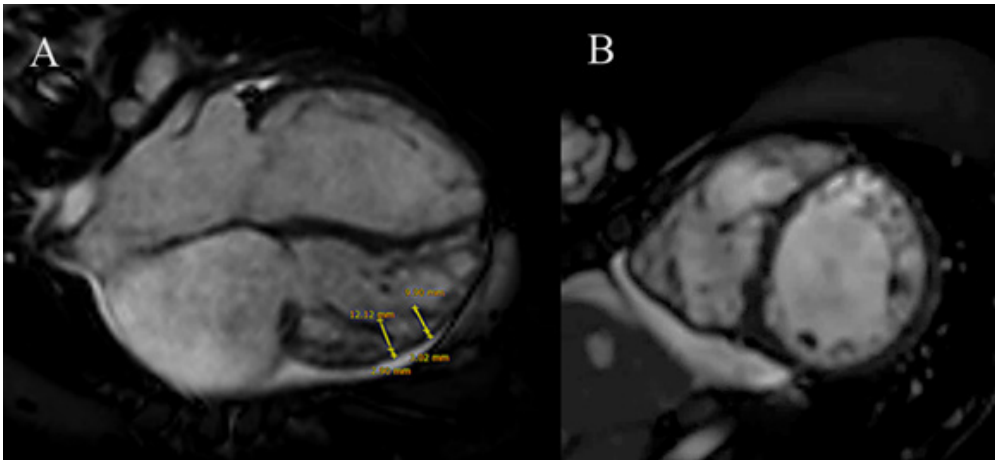


**Figure 3 —** *CMR demonstrating ventricular hypertrabeculation. A) Four-chamber view demonstrating a noncompacted-to-compacted myocardium ratio of 4.17, exceeding the cutoff value of 2.3 established by the Petersen criterion. B) Short-axis view demonstrating prominent LV myocardial trabeculation.*

## Discussion

Ventricular hypertrabeculation predominantly affects the LV, whereas biventricular involvement is less common and is generally associated with greater clinical complexity and a higher incidence of adverse outcomes. In the present case, cardiac chamber dilatation, FVR, and evidence of RV dysfunction underscore the phenotypic heterogeneity of the disease and the potential for a more severe clinical course when both ventricles are involved.[5,6]

Cardiac chamber dilatation in ventricular hypertrabeculation may lead to FVR, particularly MR and TR, as observed in this patient. This mechanism is driven by

annular dilatation and distortion of ventricular geometry, contributing to worsening HF symptoms and an increased risk of clinical decompensation.[6]

Patients with ventricular hypertrabeculation have a high prevalence of arrhythmias and cardiac conduction abnormalities, including AF and advanced AV block. The need for permanent pacemaker implantation at a young age, together with permanent AF, highlights the arrhythmogenic potential of this condition and the impact of myocardial remodeling on the cardiac conduction system.[7,8]

The American College of Medical Genetics and Genomics recommends genetic evaluation in patients with cardiomyopathies, particularly in the presence of early disease onset, an arrhythmic phenotype, or concomitant neuromuscular disorders.[9] Ventricular hypertrabeculation has been associated with muscular dystrophies, myotonic dystrophy, EDMD, and mitochondrial myopathies. In the present case, the patient remains under etiological investigation for her neuromuscular disorder, and G-banded karyotyping has not identified any chromosomal abnormalities to date.[10]

Biventricular hypertrabeculation is a rare manifestation of the disease. Previous reports have described adults presenting with HF and a family history suggestive of cardiomyopathy, patients in whom the diagnosis was established only after CMR, and young individuals presenting with severe arrhythmias or initially noncardiac manifestations.[11-13] These findings highlight the marked phenotypic variability of biventricular involvement, its substantial arrhythmogenic potential, and the pivotal role of advanced cardiac imaging in establishing the diagnosis.

## Conclusion

This case highlights the diagnostic complexity of biventricular hypertrabeculation and its association with neuromuscular manifestations, emphasizing the importance of a comprehensive etiological investigation and a multidisciplinary approach. Early recognition of the disease and follow-up at specialized centers are essential to optimize treatment, facilitate monitoring for disease-related complications, and expand our understanding of the clinical spectrum of this condition, particularly in patients with neuromuscular disorders that remain under etiological investigation.

## Author Contributions

Conception and design of the research: Gonçalo MO, Couceiro KN, Ferreira JMBB; acquisition of data: Gomes AAS, Monteiro MM, Cavalcante NB, Bezerra FA; analysis and interpretation of the data: Machado MA, Miranda DCC, Couceiro KN, Ferreira JMBB; writing of the manuscript: Gonçalo MO, Monteiro MM; critical revision of the manuscript for intellectual content: Couceiro KN, Ferreira JMBB.

### Potential Conflict of Interest

No potential conflict of interest relevant to this article was reported.

### Sources of Funding

There were no external funding sources for this study.

### Study Association

This study is not associated with any thesis or dissertation work.

### Ethics Approval and Consent to Participate

This study was approved by the Ethics Committee of the Universidade do Estado do Amazonas under protocol number 7.106.271. All the procedures in this study were in accordance with the 1975 Helsinki Declaration, updated in 2013. Informed consent was obtained from all participants included in the study.

### Use of Artificial Intelligence

The authors did not use any artificial intelligence tools in the development of this work.

### Availability of Research Data

The underlying content of the research text is contained within the manuscript.

## References

1. Chin TK, Perloff JK, Williams RG, Jue K, Mohrmann R. Isolated Noncompaction of Left Ventricular Myocardium. A Study of Eight Cases. Circulation. 1990;82(2):507-13. doi: 10.1161/01.cir.82.2.507.
2. Rosa LV, Salemi VM, Alexandre LM, Mady C. Noncompaction Cardiomyopathy: A Current View. Arq Bras Cardiol. 2011;97(1):e13-9. doi: 10.1590/s0066-782x2011000900021.
3. Finsterer J, Stöllberger C, Towbin JA. Left Ventricular Noncompaction Cardiomyopathy: Cardiac, Neuromuscular, and Genetic Factors. Nat Rev Cardiol. 2017;14(4):224-37. doi: 10.1038/nrcardio.2016.207.
4. Petersen SE, Selvanayagam JB, Wiesmann F, Robson MD, Francis JM, Anderson RH, et al. Left Ventricular non-Compaction: Insights from Cardiovascular Magnetic Resonance Imaging. J Am Coll Cardiol. 2005;46(1):101-5. doi: 10.1016/j.jacc.2005.03.045.
5. Oechslin EN, Jost CHA, Rojas JR, Kaufmann PA, Jenni R. Long-Term Follow-Up of 34 Adults with Isolated Left Ventricular Noncompaction: A Distinct Cardiomyopathy with Poor Prognosis. J Am Coll Cardiol. 2000;36(2):493-500. doi: 10.1016/s0735-1097(00)00755-5.
6. Arbustini E, Favalli V, Narula N, Serio A, Grasso M. Left Ventricular Noncompaction: A Distinct Genetic Cardiomyopathy? J Am Coll Cardiol. 2016;68(9):949-66. doi: 10.1016/j.jacc.2016.05.096.
7. Stöllberger C, Finsterer J, Blazek G. Left Ventricular Hypertrabeculation/Noncompaction and Association with Additional Cardiac Abnormalities

and Neuromuscular Disorders. Am J Cardiol. 2002;90(8):899-902. doi: 10.1016/s0002-9149(02)02723-6.

8. Maron BJ, Towbin JA, Thiene G, Antzelevitch C, Corrado D, Arnett D, et al. Contemporary Definitions and Classification of the Cardiomyopathies: An American Heart Association Scientific Statement from the Council on Clinical Cardiology, Heart Failure and Transplantation Committee; Quality of Care and Outcomes Research and Functional Genomics and Translational Biology Interdisciplinary Working Groups; and Council on Epidemiology and Prevention. Circulation. 2006;113(14):1807-16. doi: 10.1161/CIRCULATIONAHA.106.174287.

9. Hershberger RE, Givertz MM, Ho CY, Judge DP, Kantor PF, McBride KL, et al. Correction: Genetic Evaluation of Cardiomyopathy: A Clinical Practice Resource of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2019;21(10):2406-9. doi: 10.1038/s41436-019-0521-2.

10. Finsterer J, Stöllberger C, Blazek G. Neuromuscular Implications in Left Ventricular Hypertrabeculation/Noncompaction. Int J Cardiol. 2006;110(3):288-300. doi: 10.1016/j.ijcard.2005.10.028.

11. Odiete O, Nagendra R, Lawson MA, Okafor H. Biventricular Noncompaction Cardiomyopathy in a Patient Presenting with New Onset Seizure: Case Report. Case Rep Cardiol. 2012;2012:924865. doi: 10.1155/2012/924865.

12. Syed MP, Doshi A, Pandey D, Kate Y, Harizi R. A Rare Case of Biventricular Non-Compaction. BMJ Case Rep. 2020;13(5):e231154. doi: 10.1136/bcr-2019-231154.

13. Angiras S, Boruah DK, Bharadwaj R, Phukan P, Sarma K. Biventricular Noncompaction Cardiomyopathy: Rare Case with MRI Diagnosis Insights. Indian J Radiol Imaging. 2025;36(1):124-7. doi: 10.1055/s-0045-1809624.