

EDITORIAL COMMENT

Saw-Tooth Cardiomyopathy

Try Not to Stumble Twice Over the Same Stone*

Pablo Garcia-Pavia, MD, PhD,^{a,b,c,d} Fernando Dominguez, MD, PhD^{a,b,c,e}



In 1984, Engberding et al. (1) reported for the first time a case of a rare myocardial anomaly, consisting of isolated myocardial sinusoids in the absence of any other structural abnormalities. Some years later, Chin et al. (2) published a series of 8 cases and introduced the term *isolated noncompaction of left ventricular myocardium* (2), which was later modified to *left ventricular noncompaction* (LVNC). The American Heart Association considers LVNC a primary cardiomyopathy (3), whereas the European Society of Cardiology includes it in the group of unclassified cardiomyopathies because of the lack of genetic specificity and the overlap with other cardiomyopathies (4).

Until recently, the predominant pathophysiological explanation for LVNC was the embryogenic theory—which assumes that this myocardial appearance derives from an arrest of the normal compaction process of the myocardium during fetal development (5).

However, there are data supporting that LVNC could be an acquired morphological trait present in other clinical entities such as dilated cardiomyopathy, or even in the general population, and not a

distinct cardiomyopathy. For instance, it has been shown that hypertrabeculation can be acquired and is reversible in different populations, including pregnant women (6), athletes (7), or people with sickle cell anemia (8). Moreover, almost 10% of healthy individuals meet cardiac magnetic resonance imaging Petersen criteria for LVNC in 2 or more segments of the myocardium (9), and several definitions of LVNC have been proposed, reflecting the difficulty of defining LVNC as a cardiomyopathy (10).

In 2009, the first case of saw-tooth cardiomyopathy, a type of “LVNC to the extreme,” was reported in a 2-month-old infant. In that case, however, the myocardium appeared compacted, with dramatic cross-bridging muscular projections. Additionally, left ventricular ejection fraction (LVEF) was impaired, and there was an apical aneurysm and a patent foramen ovale (11). Two additional cases have also reported of concomitant LVEF impairment (12,13), which raises the question of whether hypokinetic nondilated cardiomyopathy (as an incipient form of dilated cardiomyopathy) is the main phenotype and the saw-tooth appearance is a morphologic trait.

In this issue of *JACC: Case Reports*, Proukhnitzky et al. (14) present a nicely illustrated case of saw-tooth myocardium. Unlike the previous cases, data regarding family history and genetic analysis are provided. Interestingly, no familial disease and no pathogenic variants were found in a large panel of genes, including those involved in LVNC phenotypes. We agree with the authors that new genes should be explored through exome or genome sequencing in this context, but the absence of genetic/familial findings supports that saw-tooth cardiomyopathy might not have a genetic basis.

Furthermore, according to the current knowledge about saw-tooth cardiomyopathy, the clinical approach with these patients should be similar to those presenting with the hypertrabeculated/LVNC

*Editorials published in *JACC: Case Reports* reflect the views of the authors and do not necessarily represent the views of *JACC: Case Reports* or the American College of Cardiology.

From the ^aHeart Failure and Inherited Cardiac Diseases Unit, Department of Cardiology, Hospital Universitario Puerta de Hierro, Madrid, Spain; ^bCentro de Investigación Biomédica en Red en Enfermedades Cardiovasculares, Madrid, Spain; ^cEuropean Reference Network for Rare and Low Prevalence Complex Diseases of the Heart; ^dUniversidad Francisco de Vitoria, Pozuelo de Alarcón, Spain; and the ^eMyocardial Biology Program, Centro Nacional de Investigaciones Cardiovasculares, Madrid, Spain. Both authors have reported that they have no relationships relevant to the contents of this paper to disclose.

The authors attest they are in compliance with human studies committees and animal welfare regulations of the authors' institutions and Food and Drug Administration guidelines, including patient consent where appropriate. For more information, visit the *JACC: Case Reports* [author instructions page](#).

spectrum with preserved LVEF, and only periodic cardiac surveillance is recommended. Thus, in our opinion, both entities should be generally considered as morphological traits or consequences of ventricular remodeling in the context of other cardiomyopathies but not a distinct cardiomyopathy per se (apart from very selected cases such as infantile tafazzinopathies in LVNC) (15). Accordingly, and until more information is obtained, we would recommend using the term *saw-tooth myocardium* instead of *saw-tooth cardiomyopathy*, to avoid the problems caused by LVNC terminology.

As in LVNC, we recommend focusing on LV function, treating those patients with systolic impairment according to clinical guidelines and searching for other signs of cardiomyopathy in those with normal function (e.g., arrhythmia, syncope, family history, etc.) (10,16). If LVEF is preserved and no other symptoms or signs are present, we could be facing a benign morphological trait, as in most cases of hypertrabeculation.

Although the reported case presents normal LV function, additional signs of incipient cardiomyopathy (electrocardiography with conduction defect, inferior and inferoseptal akinesia, linear intramyocardial late enhancement) are present, and as such, progression to overt LVEF dysfunction would not be a surprise.

The saw-tooth appearance of the myocardium may not meet LVNC criteria, but morphological nuances aside, we can take advantage of the lessons learned over the years with LVNC and try not to stumble twice over the same stone.

ADDRESS FOR CORRESPONDENCE: Dr. Pablo Garcia-Pavia, Heart Failure and Inherited Cardiac Diseases Unit, Department of Cardiology, Hospital Universitario Puerta de Hierro, Manuel de Falla, 2, 28222 Madrid, Spain. E-mail: pablogpavia@yahoo.es. Twitter: [@dr_pavia](https://twitter.com/dr_pavia).

REFERENCES

- Engberding R, Bender F. Identification of a rare congenital anomaly of the myocardium by two-dimensional echocardiography: persistence of isolated myocardial sinusoids. *Am J Cardiol* 1984; 1733-4.
- Chin TK, Perloff JK, Williams RG, Jue K, Mohrmann R. Isolated noncompaction of left ventricular myocardium. A study of eight cases. *Circulation* 1990;82:507-13.
- Maron BJ, Towbin JA, Thiene G, 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;113:1807-16.
- Elliott P, Andersson B, Arbustini E, et al. Classification of the cardiomyopathies: a position statement from the European Society of Cardiology Working Group on Myocardial and Pericardial Diseases. *Eur Heart J* 2008;29:270-6.
- Varnava AM. Isolated left ventricular non-compaction: a distinct cardiomyopathy? *Heart* 2001;86:599-600.
- Gati S, Papadakis M, Papamichael ND, et al. Reversible de novo left ventricular trabeculations in pregnant women: implications for the diagnosis of left ventricular noncompaction in low-risk populations. *Circulation* 2014;130: 1733-4.
- Gati S, Chandra N, Bennett RL, et al. Increased left ventricular trabeculation in highly trained athletes: do we need more stringent criteria for the diagnosis of left ventricular non-compaction in athletes? *Heart* 2013;99:401-8.
- Gati S, Papadakis M, Van Niekerk N, Reed M, Yeghen T, Sharma S. Increased left ventricular trabeculation in individuals with sickle cell anaemia: physiology or pathology? *Int J Cardiol* 2013;168:1658-60.
- Zemrak F, Ahlman MA, Captur G, et al. The relationship of left ventricular trabeculation to ventricular function and structure over a 9.5-year follow-up: the MESA study. *J Am Coll Cardiol* 2014;64:1971-80.
- Garcia-Pavia P, de la Pompa JL. Left ventricular non-compaction: a genetic cardiomyopathy looking for diagnostic criteria. *J Am Coll Cardiol* 2014;64:1981-3.
- Davlouros P, Danias P, Karatza A, Kiaffas M, Alexopoulos D. Saw-tooth cardiomyopathy. *J Cardiovasc Magn Reson* 2009;16:54.
- de Pinho Cardoso B, Trigo C, Jalles Tavares N, Pinto FF. Sawtooth cardiomyopathy: a rare cause of heart failure. *Rev Port Cardiol* 2017;36:875-6.
- Chenaghoulou M, Kasaei M, Taghavi S, Amin A, Naderi N. Saw tooth cardiomyopathy: a case report. *ESC Heart Fail* 2020;7:325-8.
- Proukhnitzky J, Garot J, Bordet C, et al. Sawtooth cardiomyopathy clinical presentation and genetic analysis. *J Am Coll Cardiol Case Rep* 2020; 2:1205-9.
- Arbustini E, Favalli V, Narula N, Serio A, Grasso M. Left ventricular noncompaction: a distinct genetic cardiomyopathy? *J Am Coll Cardiol* 2016;68:949-66.
- Aung N, Zemrak F, Mohiddin SA, Petersen SE. LV noncompaction cardiomyopathy or just a lot of trabeculations? *J Am Coll Cardiol Img* 2017;10:704-7.

KEY WORDS cardiac magnetic resonance, cardiomyopathy, dysplasia, echocardiography, fibrosis, genetic disorders, left ventricle, phenotype