CASE REPORT

Folia Cardiologica 2022
vol. 17, no. 1, pages 62–64
DOI: 10.5603/FC.2022.0009

ISSN 2353–7752
e-ISSN 2353–7760

Left ventricular non-compaction cardiomyopathy: a rare cause of cardiovascular complaints

Azita Rezaei[1], Marcin Gregorczyk[1], Agnieszka Ciba-Stemplewska[2], Katarzyna Starzyk[3], Wioletta Korzeluch[4], Michał Spałek[5, 6], Beata Wożakowska-Kapłon[1, 3]

[1]Faculty of Medicine and Health Sciences, *Collegium Medicum*, Jan Kochanowski University in Kielce, Poland
[2]Department of Internal Medicine, Regional Polyclinical Hospital in Kielce, Poland
[3]1st Department of Cardiology and Electrotherapy, Świętokrzyskie Cardiology Centre, Regional Polyclinical Hospital in Kielce, Poland
[4]Cardiology Outpatient Clinic, Świętokrzyskie Paediatric Centre, Regional Polyclinical Hospital in Kielce, Poland
[5]Institute of Anatomy, Collegium Medicum, Jan Kochanowski University in Kielce, Poland
[6]Institute of Diagnostic Imaging, Holy Cross Cancer Centre in Kielce, Poland

Abstract

20-year-old woman with diagnosed in childhood left ventricular non-compaction (LVNC) cardiomyopathy with coexisting arthrogryposis was admitted to the Department of Cardiology due to atypical chest pain, worsening of exercise tolerance, bendopnea and symptoms of bronchial asthma exacerbation. The electrocardiogram (ECG) showed changes indicative of ischemia, without their dynamics. Inflammatory parameters, BNP (B-type natriuretic peptide), and myocardial necrosis markers were not elevated. Systolic and diastolic function on echocardiography (ECHO) was assessed as normal. Despite the initially suggestive clinical picture, the correct direction of the differential diagnosis, and consequently the diagnosis, was established after completing the history of comorbidities.

Key words: LVNC, cardiomyopathy, arthrogryposis

Folia Cardiologica 2022; 17, 1: 62–64

Introduction

Left ventricular non-compaction cardiomyopathy (LVNC) is a rare congenital disease that can occur familially and spontaneously [1]. The prevalence is 0.05–0.26% of all adults referred for echocardiography and 3–4% of patients with heart failure (HF) [2].

During the embryonic period, the myocardium is a trabecular structure, supplied with blood in the intertrabecular spaces called sinusoids (recesses). With the development of coronary circulation in the pericardium, the sinusoids disappear and the walls of the ventricles transform into a compact muscle. This process proceeds beginning from the base of the heart and ending at its apex. In LVNC, this process is disrupted. As a result, hearts with LVNC are characterised by excessive trabeculation and deep intertrabecular sinusoids. Although coronary circulation is normal, wall structure abnormalities may adversely affect myocardial wall perfusion, causing ischaemia and leading to thromboembolic incidents, arrhythmias, or heart failure [3]. Treatment consists mainly of preventing these complications. Genetic mutations are cited as the cause, with 60% of them being spontaneous. The rest of the cases are autosomal

Address for correspondence: Azita Rezaei, Wydział Lekarski i Nauk o Zdrowiu, *Collegium Medicum,* Uniwersytet Jana Kochanowskiego w Kielcach, aleja IX Wieków Kielc 19A, 25–317 Kielce, Poland, e-mail: azita.rezaei91@gmail.com

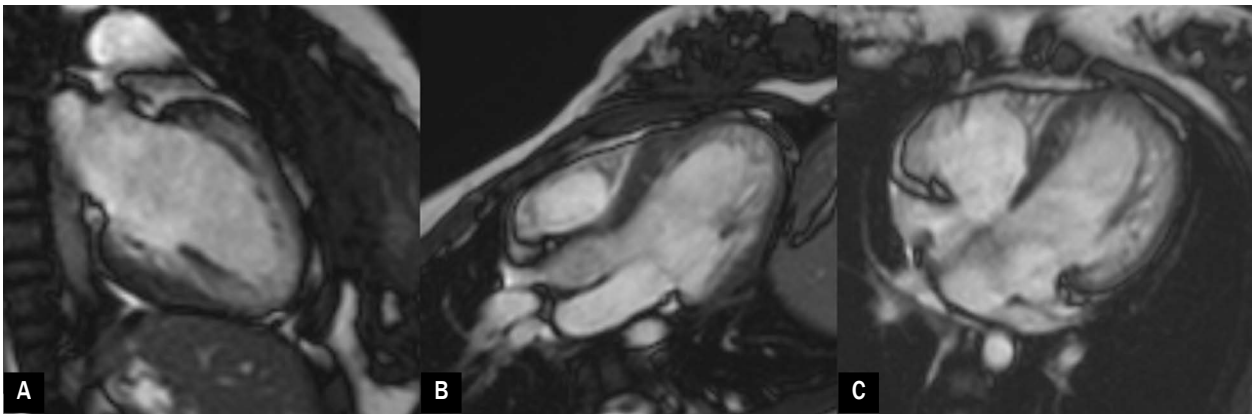

Figure 1. An magnetic resonance image showing increased trabeculation; **A.** 2-chamber view; **B.** 3-chamber view; **C.** 4-chamber view

dominant mutations with incomplete penetrance or coupled to the X chromosome (e.g. Barth syndrome) [2]. Mutations affect sarcomeric genes that are common causes of hypertrophic and dilated cardiomyopathy: *MYH7*, *MYBPC3*, *TTN*.

There are four subtypes of LVNC, provided that the dimensions of the left ventricle (LV) are known:

- isolated LVNC (if LV dimensions are normal, without hypertrophy);
- LVNC with hypertrophic cardiomyopathy (if LV wall thickness is ≥ 13 mm, or there was a previous diagnosis of hypertrophy);
- LVNC with dilated cardiomyopathy (if there is an enlargement of LV);
- LVNC with hypertrophic and dilated cardiomyopathy (when there are features of both increased LV wall thickness and enlargement of LV) [4].

The main tools in the diagnosis of LVNC are echocardiography and magnetic resonance imaging (MRI) (Figure 1). These methods enable the detection of the presence of two layers of the myocardium: a thin — compact (outer) layer and a thicker — non-compact (inner) layer, located mainly in the LV apex and the middle segments of the inferior and lateral wall. In addition, there is increased LV trabeculation with deepening of the intertrabecular sinuses, deeply perfused as seen on colour Doppler.

The most commonly used are the echocardiographic criteria proposed by Jenni et al. [5]:

- a characteristic image of multiple, excessive trabeculation with deep intertrabecular folds;
- intertrabecular spaces filled with blood flowing directly from LV cavity on colour Doppler imaging;
- a ratio of non-compacted to compacted layer thickness higher than 2 (measured in end-systolic phase, parasternal short axis view);
- no other cardiac anomalies.

The diagnostic criteria in MR are also based on calculating the ratio of non-compacted to compacted layer thickness with a cut-off point ≥ 2.3 for diagnosis [2].

Case study

A 20-year-old woman was admitted to the Cardiology Department due to exertional dyspnoea, weakness, fever (up to 38.8°), reduced exercise capacity corresponding to New York Heart Association (NYHA) class II/III along with stabbing chest pain for several days. The patient had comorbidities: bronchial asthma and arthrogryposis (a congenital multi-joint contracture disease). The patient had a history of pain and moderate exertional dyspnoea due to comorbidities.

The diagnosis of LVNC was made at the age of fourteen when she presented to her doctor because of reduced exercise tolerance and chest pain. At that time, ECG showed ST-segment depression, while echocardiogram revealed trace tricuspid regurgitation with right ventricle–right atrium (RV–RA) gradient, concentric LV hypertrophy with hypertrophied endocardium, right ventricular apical hypertrophy, markedly reduced contractility of the basal region. The family history was negative.

Laboratory tests were performed on admission. Inflammatory markers, markers of myocardial necrosis, and BNP were all within the normal range.

An ECG was performed which showed: sinus rhythm, J-point elevation in V1–V3 leads, with flattened T-waves in I, II, aVL, and V5–V6 leads (Figure 2). Neither the dynamics of ECG changes nor elevated levels of markers of myocardial necrosis were observed.

No significant LV systolic dysfunction or segmental wall motion abnormalities were observed on the echocardiogram. The only abnormality was the increased trabeculation characteristic of LVNC.

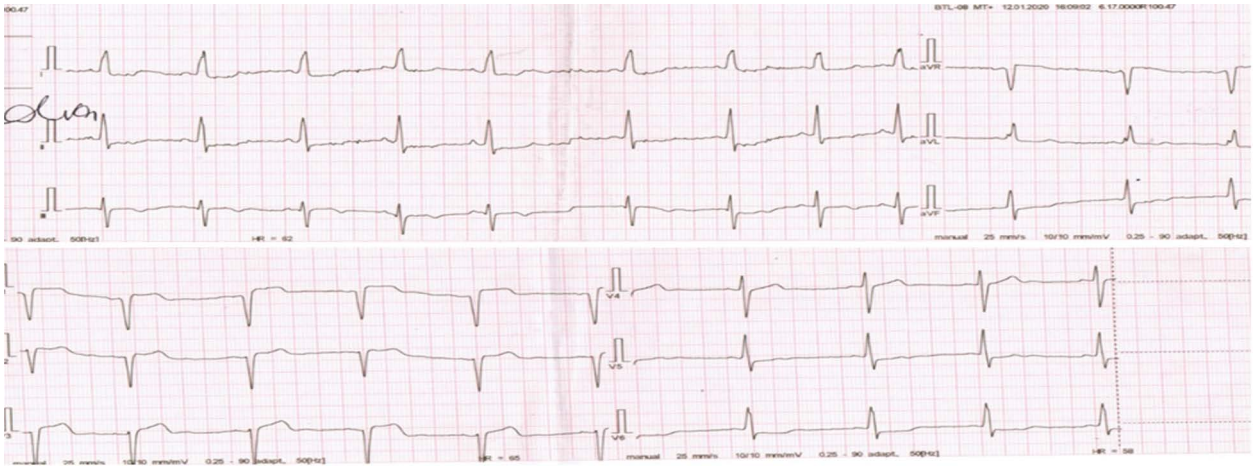

Figure 2. Electrocardiogram on admission: sinus rhythm, J-point elevation in V1–V3 leads, with flattened T-waves in I, II, aVL, aVF, and V5–V6 leads

Discussion

The case described reflects the difficulties in differential diagnosis and the management of treatment. Patients with LVNC, as in this case, may have persistent ECG changes. Therefore, in order to rule out newly developed disorders, results should be compared with previous ones, if available. Likewise, heart failure, common in LVNC, was ruled out by echocardiography and by normal BNP levels.

Other causes of reported complaints were therefore taken into account:

— infectious exacerbation of bronchial asthma (increased spasticity over the lung fields, fever);
— pain associated with musculo-articular disorders.

Conclusions

LVNC, like arthrogryposis, is a rare disease. LVNC may present individually differentiated ECG changes: features of LV or left atrial hypertrophy, ventricular repolarisation abnormalities, signs of preexcitation. Dyspnoea may result from LV systolic dysfunction, arrhythmias, thromboembolic complications. ECG and echocardiography, due to their availability, are fundamental in establishing the correct diagnostic procedure. If new symptoms appear, the medical history should also be carefully taken and co-morbidities should be taken into account. In the described patient, despite a diagnosis suggestive of rare cardiomyopathy, her complaints were caused by an infectious exacerbation of bronchial asthma and musculoskeletal disorders.

Conflict of interest

The authors declare no conflicts of interest.

Funding

None.

References

1. Singh DP, Patel H. Left ventricular non-compaction cardiomiopathy. StatPearls Publishing 2021. https://www.ncbi.nlm.nih.gov/books/NBK537025/ (July 10, 2021).
2. Ponikowski P, Hoffman P, Witkowski A. ed. Kardiologia. Podręcznik Polskiego Towarzystwa Kardiologicznego. Via Medica, Gdańsk 2019: 478, 488.
3. Ichida F. Left ventricular noncompaction — risk stratification and genetic consideration. J Cardiol. 2020; 75(1): 1–9, doi: 10.1016/j.jjcc.2019.09.011, indexed in Pubmed: 31629663.
4. van Waning JI, Moesker J, Heijsman D, et al. Systematic review of genotype-phenotype correlations in noncompaction cardiomyopathy. J Am Heart Assoc. 2019; 8(23): e012993, doi: 10.1161/JAHA.119.012993, indexed in Pubmed: 31771441.
5. Oechslin E, Jenni R. Left ventricular non-compaction revisited: a distinct phenotype with genetic heterogeneity? Eur Heart J. 2011; 32(12): 1446–1456, doi: 10.1093/eurheartj/ehq508, indexed in Pubmed: 21285074.