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CASE REPORT

A common pediatric presentation of an unusual disease: non-compaction cardiomyopathy

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ABSTRACT

Background: Shortness of breath is a very common presentation at the pediatric emergency department, but making a diagnosis based on it is very challenging for physicians because of the numerous differential diagnoses, including respiratory and cardiac causes.

Case Presentation: Herein, we describe the case of a 2-month-old patient who presented to our emergency department in October 2017 with shortness of breath for 3 weeks duration. She was diagnosed as having lower respiratory tract infection; a cardiac murmur was incidentally found, which turned out to be a very rare congenital cardiac disease: left ventricular non-compaction cardiomyopathy (LVNC). She was treated with anti-failure medications but unfortunately died before cardiac transplant.

Conclusion: As demonstrated by this case, non-compaction cardiomyopathy can be easily missed and misdiagnosed because of its rarity and diverse symptoms on presentation. Since corresponding treatment modalities are lacking, the prevention of associated complications is the primary goal of LVNC management.

Keywords: Pediatric, pediatric emergency, cardiology, cardiomyopathy.

Background

Despite an increasing awareness and interest in non-compaction cardiomyopathy (NCC), information on the diagnostic criteria, symptoms, and prognosis of this rare and unique congenital disorder is limited. Herein, we describe the case of a patient who had a common pediatric presentation and was incidentally found to have NCC.

Case Presentation

A 2-month-old girl part of a twin, 34-week pregnancy was born via normal vaginal delivery with a birth weight of 2.2 kg. She remained in hospital for 5 days due to neonatal jaundice that required only phototherapy. She was brought to our emergency department (ED) with a reported history of tachypnoea for the 3 weeks prior to the current presentation, which was noticed by her parents in comparison with her twin sister's condition. The tachypnoea gradually became severe and was associated with a non-productive cough, nasal congestion, and interrupted feeding for 3 days preceding the current presentation. There was no history of cyanosis, sweating with feeding, or decreased urine output. The parents initially sought medical advice from a pediatric clinic, and the patient was diagnosed with bronchiolitis. Her symptoms worsened over subsequent weeks, however, prompting them to bring her to the ED.

On examination, the patient was in respiratory distress, as indicated by nasal flaring and subcostal retractions. She did not have cyanosis or dysmorphic features. Her respiratory rate was 64 breaths/minute (above the 95th percentile for age), heart rate was 174 beats/minute (above the 95th percentile for age), and oxygen saturation was 100% on room air. Four-limb blood pressure readings were all just below the 50th percentile for age.

Cardiovascular examination revealed palpable equal peripheral pulses with no central – peripheral delays, a quiet pericardium, and no visible pericardial pulsation. The first and second heart sounds were audible, with a soft systolic murmur (grade 3/6) mainly in the left sternal border. Her chest had good air entry bilaterally, with transmitted sounds and no wheezing. The abdomen was

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soft, with a palpable liver edge located approximately 2 cm below the right costal margin. A chest X-ray was obtained and showed cardiac enlargement and lung parenchymal disease (Figure 1) along with an electrocardiogram (ECG) (Figure 2) that was normal.

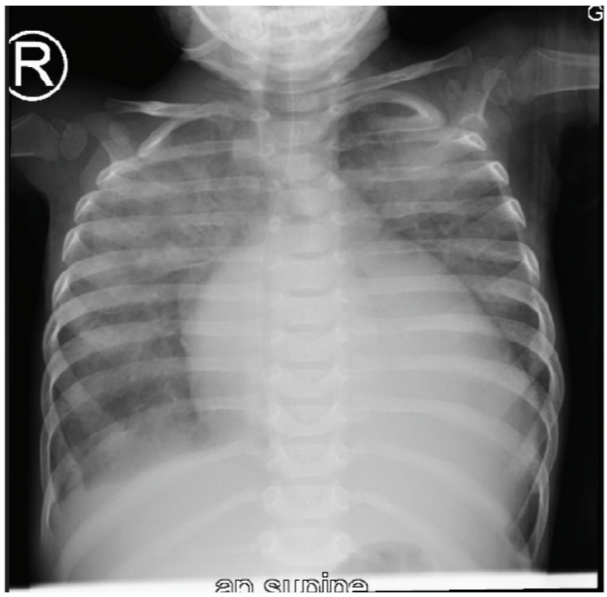

Figure 1. *Chest x-ray.*

Cardiology referral and echocardiography (Figure 3) were done in the ED. The examination revealed a small ostium secundum atrial septal defect shunting from left to right, and left ventricular non -compaction cardiomyopathy (LVNC) (4.8%), with fair right ventricular function and very poor left ventricular function with an ejection fraction of 10%. During admission, her twin sister also underwent echocardiography screening, and she exhibited a normal heart structure. The patient and her twin sister also underwent genetic testing, but the results were not available in the patient's file.

Respiratory distress is one of the most common presentations in the pediatric ED. Recognizing respiratory distress is straightforward, but identifying its causes can be challenging because the differential diagnosis varies and includes respiratory, cardiac, metabolic, and central nervous system conditions. Respiratory distress accounts for more than 9 million pediatric ED visits in the United States. It also accounts for 11% of all pediatric ED visits and 25% of all pediatric hospital admissions [1].

Primary respiratory pathologies, i.e., pneumonia and bronchiolitis were not at the top of the list in the differential diagnosis because the patient lacked a fever and had endured a long period of illness (3 weeks). A cardiac pathology was considered because of the chronicity of the illness, the patient's age, and the presence of tachycardia,

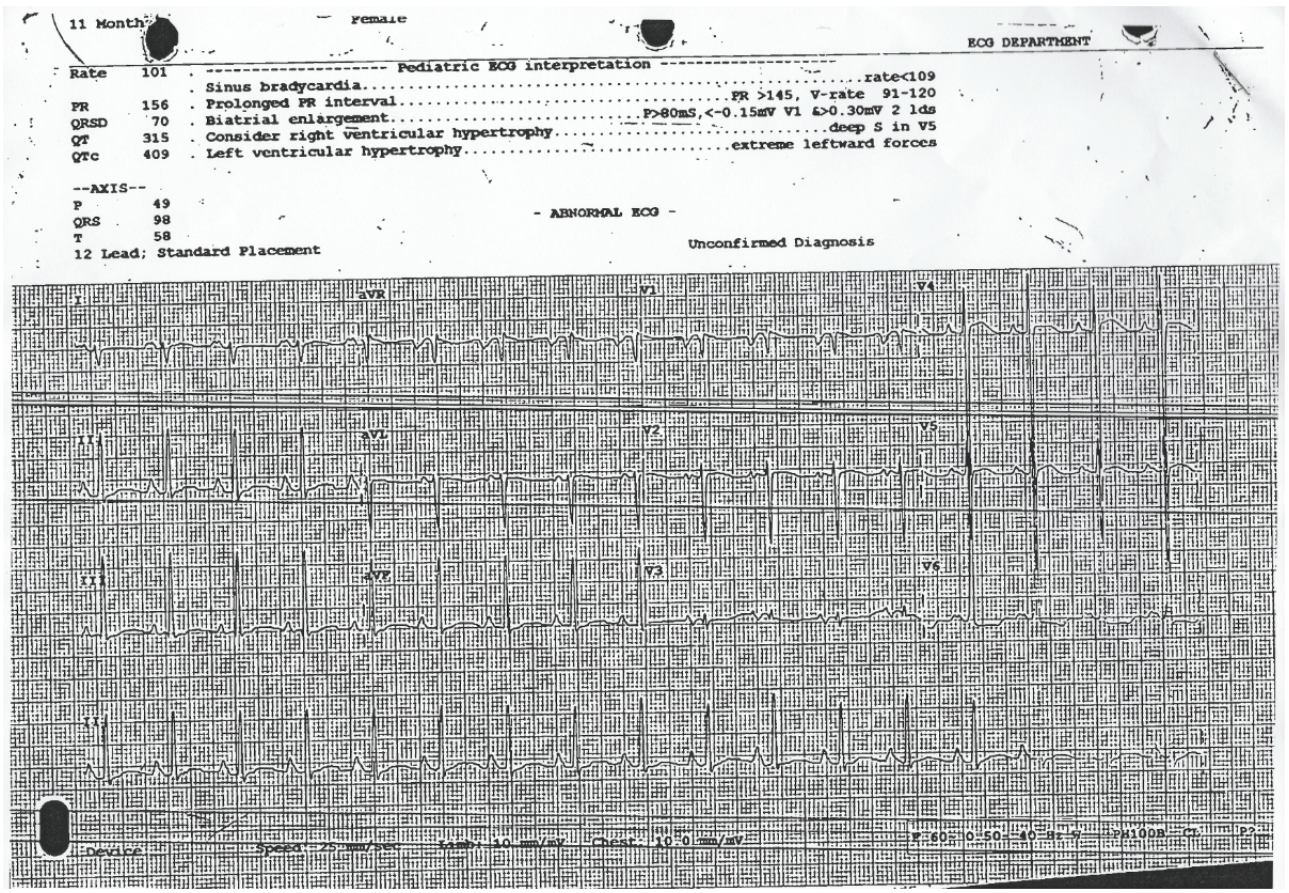

Figure 2. *ECG.*

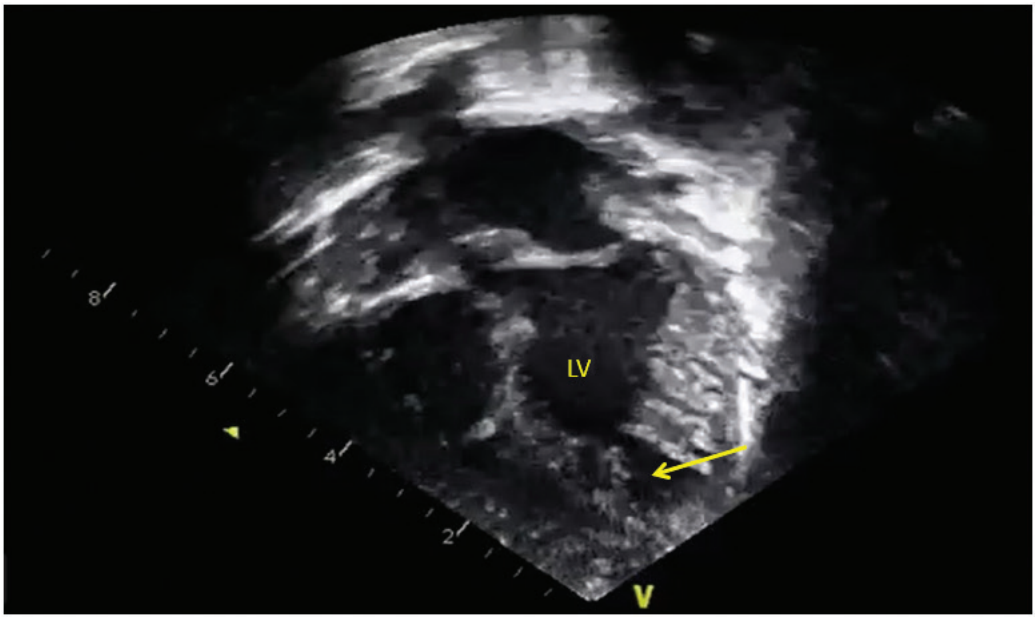

Figure 3. *Echo showing the trabeculation.*

murmur, and hepatomegaly. Based on the patient's general appearance, normal oxygen saturation, and four-limb blood pressure measurements, the initial suspicion was non-cyanotic heart disease or coarctation of the aorta, which are the most common congenital cardiac anomalies observed in the ED. Instead, we favored a diagnosis of heart failure secondary to congenital heart disease. Based on that diagnosis, an urgent cardiac consultation was performed in the ED.

The patient was admitted to the cardiac intensive care unit (ICU) where inotropes were started initially along with anti-failure medications (furosemide and captopril) and aspirin.

The patient was discharged 5 days later in a fair condition. She was followed up in the clinic for five months with several admissions to the pediatric ward and ICU because of heart failure. Unfortunately, she died before cardiac transplant.

Discussion

LVNC is a rare congenital cardiac disorder that was first reported and described pathologically by Grant in 1926 [1,2]. The American Heart Association recognized LVNC as a distinct type of cardiomyopathy in 2006 [3]. It was previously called spongy myocardium or hypertrabeculation syndrome, but these terms should not be used interchangeably with reference to LVNC [1].

LVNC was thought to be caused by an arrest in the compaction process that occurs between weeks 5 and 8 of gestation and involves a loose meshwork of the cardiac muscle fibers that link the myocardial wall with the ventricular cavity. Notably, however, the precise mechanisms involved remain unclear given the wide age range in which the disease manifests. According to Ritter et al. [4], LVNC is characterized by (a) an altered myocardial wall with prominent trabeculae and deep intertrabecular recesses, resulting in thickened myocardium with two layers consisting of non-compacted myocardium and a thin, compacted layer of myocardium; and (b) continuity between the deep intertrabecular recesses and the left ventricular cavity, from which it is filled with blood [4,5].

LVNC can occur as an isolated cardiac anomaly. Such cases are termed isolated LVNC, and reportedly occurred at an estimated rate of 0.014% to 1.3% in patients who underwent echocardiography [6], but the prevalence of the condition in the general population is unknown. LVNC can also occur in association with other cardiac anomalies such as patent ductus arteriosus, patent foramen ovale, tricuspid atresia, right ventricular hypoplasia, Ebstein anomaly, bicuspid aortic valve, and transposition of the great arteries. Atrial septal defect, ventricular septal defect, or fistulas of the right coronary artery may also be present according to a retrospective study conducted by Ergul et al. [7]. Isolated LVNC and non-isolated LVNC have been reported to be associated with several electrocardiographic changes such as ST depression, flat or negative T waves, bundle branch block, Wolff–Parkinson–White syndrome, and numerous patterns of arrhythmia. Ogawa et al. [8] recently reported two cases of LVNC associated with long QT syndrome.

More cases of LVNC are being discovered lately due to increased awareness of the condition and improved imaging techniques that yield greater resolution of LVNC trabeculations. LVNC can be sporadic or familial. The sporadic causes of NCC are unclear and an estimated 86% of cases are idiopathic [9]. Jefferies et al. [9] reported

that the remaining non-idiopathic diagnoses were due to metabolic diseases (including Barth syndrome, Charcot–Marie–Tooth disease, and Melnick–Needles syndrome), malformation syndrome, or myocarditis. The familial type of NCC is more commonly transmitted via autosomal dominant inheritance rather than X-linked or autosomal recessive inheritance, as suggested by Zaragoza et al. [10].

The age at clinical presentation of LVNC varies, with some patients diagnosed during the neonatal period and others incidentally discovered at an older age. Clinical manifestations of LVNC differ widely. Patients with LVNC can present with symptoms of heart failure, atrial or ventricular arrhythmias, or thromboembolic events such as stroke [3,11,12].

Some patients can be asymptomatic and diagnosed incidentally via abnormal findings during the cardiac examination, chest radiography, or echocardiography [4,13]. Ozgur et al. reported one patient who was diagnosed at 15 years of age during family screening [13]. Based on a small series of 16 cases, Ritter et al. [4] concluded that the average time until the onset of symptoms after diagnosis was 3.5 years. In Oechslin et al. [11] study, the clinical manifestations at the time of diagnosis included dyspnea in 27 patients (79%), heart failure in 12 (35%), chest pain in 9 (26%), and chronic atrial fibrillation in 9 (26%). Our patient presented solely with a 3-week history of dyspnea, and a cardiac murmur was detected during subsequent echocardiography. In 2013, Ergul et al. [7] described a 4-year-old boy with LVNC who presented with sudden cardiac death and was resuscitated and admitted to the ICU.

LVNC is frequently misdiagnosed as dilated cardiomyopathy, hypertrophic cardiomyopathy, restrictive cardiomyopathy, double-chambered left ventricle, or endomyocardial fibroelastosis, as observed by Ergul et al. [7]. The diagnosis of LVNC is usually established via transthoracic two-dimensional echocardiography. If the result is indeterminate, cardiovascular magnetic resonance imaging, 64-slice multidetector computed tomography, and left ventriculography can be used as additional diagnostic imaging modalities.

Other echocardiographic findings associated with LVNC include reduced global left ventricular systolic function, reduced *diastolic function*, left ventricular thrombi, and an abnormal papillary muscle structure [11]. LVNC may manifest in association with other types of cardiomyopathies. In Jefferies et al.'s study [9] of 3,219 patients with cardiomyopathy, LVNC was observed in 155 patients (4.8%) who were further sub-divided based on five phenotypic classifications; isolated LVNC (n = 35; 22.6%), LVNC with dilated cardiomyopathy (n = 91; 58.7%), LVNC with hypertrophic cardiomyopathy (n = 17; 11%), LVNC with indeterminate cardiomyopathy (n = 12; 7.7%), and LVNC with restrictive cardiomyopathy (0; 0%) [9].

Isolated LVNC is associated with high morbidity and mortality in children and adults. Outcomes are more closely related to the severity of the disease at clinical presentation rather than the diagnosis *per se*. In a single-center study in Texas, 242 children with isolated LVNC were identified, and after a median follow-up of 4 years the following were noted: the mortality rate was 12.8% (31 patients); 13 patients (5.4%) received a heart transplant; cardiac dysfunction developed in 150 patients (62.0%); electrocardiographic abnormalities, including ventricular hypertrophy and depolarization abnormalities were present in 211 patients (87.2%); arrhythmias developed in 80 patients (33.1%), of whom14 (17.4%) had ventricular tachycardia; and sudden cardiac death occurred in 15 patients (6.2%) [12].

There is no specific therapy for LVNC, therefore, the main aim is to prevent associated complications, namely progressive heart failure, arrhythmias, and thromboembolism. Patients must undergo Holter monitoring annually to monitor for asymptomatic arrhythmias, and in cases of symptomatic ventricular arrhythmias with impaired systolic function, anti-arrhythmic agents or implantable cardiac defibrillators are indicated [12]. Some authors have recommended prophylactic anticoagulation in the long term for all patients diagnosed with ventricular non-compaction, regardless of ventricular function [4,11]. Lastly, patients with LVNC may develop end-stage heart failure and require heart transplantation.

Learning Points/Take Home Messages

- NCC is a rare disease that can mimic other more common pediatric illnesses such as bronchiolitis.
- Due to its rarity and the diversity of potential symptoms on presentation, NCC can be easily missed and misdiagnosed.

List of Abbreviations

ECG	Electrocardiogram
ED	Emergency department
ICU	Intensive care unit
LVNC	Left ventricular non-compaction cardiomyopathy
NCC	Non-compaction cardiomyopathy

Conflict of Interest

The authors declare that there is no conflict of interest regarding the publication of this article.

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Consent for publication

A written consent from parent was obtained.

Ethical approval

Ethical approval is not required at our institution to publish an anonymous case report.

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