Echocardiogram versus Cardiac Magnetic Resonance Imaging for the Diagnosis of Noncompaction Cardiomyopathy – A Case Report

Sami Sameer Al Hashimi & Rami Atef Neematallah

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CASE REPORT

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1. INTRODUCTION

Left ventricular non-compaction cardiomyopathy (LVNC) is a rare disorder characterized by abnormal myocardial trabeculations and deep intertrabecular recesses. Diagnostic criteria for LVNC are controversial. Echocardiography remains the primary modality for diagnosis, but there is growing evidence that it may fail in early diagnosis due to its limitation in accurately visualizing the apical and lateral wall trabeculations. This case report describes the use of Cardiac Magnetic Resonance (CMR) imaging to diagnose early LVNC in a young patient where echocardiography had failed. The patient presented with recurrent palpitations and an unremarkable past medical history. The echocardiography results showed only mild hypokinetic changes of the apex, but more than 4,000 ventricular ectopic beats were recorded by Holter monitoring. CMR was performed to exclude myocarditis and cardiomyopathy, and non-compaction ratio measurements met the diagnostic criteria for LVNC. Treatment with a combination of a low-dose ACE inhibitor and a ß-blocker was started and the patient reported almost complete resolution of his symptoms.

Keywords: Left ventricular non-compaction cardiomyopathy, Echocardiography, Cardiac magnetic resonance imaging, Diagnosis, ß-blocker, ventricular ectopic beats.

1. INTRODUCTION

Left ventricular non-compaction cardiomyopathy (LVNC) is classified by the American Heart Association as a primary genetic cardiomyopathy. It is characterized by abnormal myocardial trabeculations and deep intertrabecular recesses, which are thought to occur as a consequence of failed compaction of the ventricular myocardium during embryogenesis [1]. Spongy myocardium was first described in 1975, however, clinical identification with two-dimensional echocardiography did not occur until 1984. LVNC is a rare disorder and reported prevalence in adults ranges from 0.014% to 0.05% with a median age at diagnosis between 40 and 50 years, predominantly in males [1 - 4]. Clinical manifestations range from asymptomatic to patients who develop palpitations, shortness of breath, heart failure, arrhythmias, thromboembolic events, to sudden death. If diagnosed late or left untreated, progressive ventricular dysfunction may require internal cardiac defibrillator implantation or lead to death.

There is controversy surrounding the diagnostic criteria for LVNC. Traditionally, it has been diagnosed by echocardiography, but in recent years, improvements in imaging tech-

2. PATIENT INFORMATION

A 31-year-old male patient was referred to the cardiology clinic with recurrent palpitations. He described palpitations with abrupt onset and termination and said they “felt like a skipped heartbeat” of very short duration, and they were accompanied by mild dizziness. They occurred on exertion almost daily but did not cause him to wake up at night or lose consciousness.

The patients’ past medical history was unremarkable, apart from asthma controlled with long-acting beta-agonists with symptoms seldom at night. The patient reported occasional gastroesophageal reflux that he treated with an oral low- dose proton-pump inhibitor. He did not take any other medications. Approximately one month prior to his presentation at the clinic, he had experienced flu-like symptoms and fever, which did not recur. He had no history of anemia or thyroid disease, past surgical history was noncontributory, and his family history was negative for heart diseases.

The patients’ social history showed that he worked in the...
oil sector and frequently drove long distances during work hours. He described his job as exhausting. He was a non-smoker, usually drank one cup of coffee per day, and did not consume energy drinks or alcohol.

3. CLINICAL FINDINGS

On physical examination, he appeared to be a healthy male, afebrile, and in no distress. His body mass index (BMI) was 33 kg/m² and his vital signs were normal. Mild expiratory wheezing was noted on chest examination. His cardiovascular examination showed normal jugular venous pressure, frequent ectopic beats, and a faint systolic murmur at the apex. The location and intensity of the apical impulse were normal.

4. DIAGNOSTIC ASSESSMENT

Routine biochemical tests revealed a slightly elevated fasting blood glucose, but glycated hemoglobin (HbA1C) was normal, and therefore, diabetes was ruled out. Thyroid panel and hemoglobin were within normal range and cardiac B-type natriuretic peptide (BNP) was normal. The electrocardiogram (ECG) was unremarkable for the exception of a few ventricular ectopic beats with the right bundle branch block (RBBB). The echocardiography also showed a normal heart shape with an intact pericardium, mild apical left ventricular (LV) hypokinesia, and an ejection fraction of 50-55%. Diastolic function was normal and there was no pulmonary hypertension. Minimal mitral regurgitation was observed without mitral valve prolapse. The patient took part in an exercise stress test to rule out coronary artery disease, during which he reached stage 5 of the Bruce Protocol without chest pain or ST-segment changes. He experienced shortness of breath at peak exercise level and demonstrated normal heart rate recovery and blood pressure response to exercise. Duke Treadmill Score was 7 (low risk) and the test was negative for inducible cardiac ischemia. Additionally, there was no exercise-induced arrhythmia. During 24-hour Holter monitoring, a total of 4384 ventricular ectopic activities were recorded, presenting as RBBB (likely originating from the left side) with couplets, bigeminy, and trigeminy. There were no triplets and no evidence of ventricular tachycardia. Heart rate variability and QT intervals were normal.

Due to the mild apical hypokinesia, a normal exercise stress test, the Holter recording of more than 500 ventricular ectopic beats per 24 hours, and a recent history of flu-like symptoms, cardiac magnetic resonance imaging (CMR) were performed to exclude myocarditis and cardiomyopathy. Long-axis and short-axis CMR diastolic non-compaction ratio measurements were recorded as follows; (*Apical long axis 4 chamber view: Diastolic compacted LV thickness 6mm, angle 66 & Diastolic non-compacted LV thickness 16 mm, angle 86; Non compacted to compacted ratio - 2.68. *Long axis 2 chamber view: Diastolic compacted thickness 5.6mm, angle 55 & Diastolic non-compacted thickness 18.0mm, angle 58; Non compacted to compacted 3.214mm) and met the generally accepted diagnostic criteria for LVNC proposed by Petersen et al.[5], LV trabeculations with a non-compacted to compacted (NC/C) ratio greater than 2.3, and changes were limited to the apical and apical lateral regions (Figs. 1 and 2). A study by Jacquier et al. [6] showed that a value of trabeculated LV mass above 20% of the global mass of the LV predicts the diagnosis of LVNC with a high sensitivity and specificity. The measured ratio of LV trabecular mass to global mass in the patient was greater than 20%.

Fig. (1). Left ventricular trabeculations, as seen on short-axis view of cardiac magnetic resonance imaging.
Fig. (2). Left ventricular trabeculations as seen on long-axis view of 4 chamber & 2 chamber cardiac Magnetic Resonance Imaging (Measurement highlighted).

Table 1. Timeline of relevant medical history and interventions.

<table>
<thead>
<tr>
<th>Date</th>
<th>Case summary</th>
<th>Diagnostic testing</th>
<th>Interventions</th>
</tr>
</thead>
<tbody>
<tr>
<td>06/06/2018</td>
<td>Patient presents with palpitations.</td>
<td>ECG, chest X-ray</td>
<td>24-hour Holter monitoring begins</td>
</tr>
<tr>
<td></td>
<td></td>
<td>CBC, TSH, electrolytes, non-fasting glucose.</td>
<td></td>
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<tr>
<td>07/06/2018</td>
<td>Patient follow-up</td>
<td>Echocardiogram. Exercise stress test.</td>
<td></td>
</tr>
<tr>
<td></td>
<td>• Routine biochemical tests are normal.</td>
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<tr>
<td></td>
<td>• ECG results suggest LVH.</td>
<td></td>
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<tr>
<td></td>
<td>• Chest X-ray is normal.</td>
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<tr>
<td>08/06/2018</td>
<td>Patient follow-up and Holter analysis.</td>
<td></td>
<td>CMR requested</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Amiodarone 200 mg PO od. Bisoprolol 1.25 mg PO od</td>
</tr>
<tr>
<td>02/12/2018</td>
<td>Patient lost to follow-up for 6 months. Still with recurrent palpitations, mild shortness of breath, does not take any medication.</td>
<td>ECG repeated: LVH is unchanged.</td>
<td>CMR (22/12/2018) Patient restarted on: Amiodarone 200 mg PO od.Bisoprolol 1.25 mg PO od.</td>
</tr>
<tr>
<td>08/01/2019</td>
<td>Patient follow-up</td>
<td></td>
<td>Detailed patient education completed.</td>
</tr>
<tr>
<td></td>
<td>• CMR result: non-compaction cardiomyopathy confirmed.</td>
<td></td>
<td>Amiodarone discontinued. Bisoprolol increased to 2.5 mg PO od. Ramipril 1.25 mg PO od added. Patient scheduled for bi-weekly follow-up visits to titrate anti-heart failure treatment.</td>
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</table>
Based on the clinical symptoms (palpitations, shortness of breath), the subtle changes observed on echocardiography, and the presence of all three CMR criteria suggested by Petersen et al., the patient was diagnosed with LVNC.

5. THERAPEUTIC INTERVENTION

The patient was started on a combination of a low-dose ACE inhibitor and a β-blocker, which was up-titrated to the maximum tolerated dose over the following weeks. Despite being asthmatic, he was able to tolerate oral bisoprolol 10 mg once daily. He reported significant improvement and almost complete resolution of his exertional symptoms and palpitations. His asthma symptoms also improved (Table 1).

6. DISCUSSION

Imaging has a pivotal role in the diagnosis of LVNC. Widely used echocardiographic diagnostic criteria for isolated LVNC include: 1) absence of coexisting cardiac abnormalities, 2) non-compaction to compaction ratio of ≥2:1 at end-systole, 3) segmental thickening of the LV myocardium with a thin compacted epicardial layer and a thick non-compacted endocardial layer with trabeculations and deep recesses, 4) color doppler evidence of deep intertrabecular recesses and 5) predominant localization in the apical, mid-lateral, and mid-inferior regions [7]. While echocardiography may provide the first clue to diagnose LVNC, there is growing evidence that it may fail in early diagnosis due to its relative difficulty to image the apex well. There are reports of delayed or missed LVNC diagnoses due to a lack of detection on echocardiography. Others have presented evidence that supports CMR over conventional echocardiography for the diagnosis of LVNC [8].

This patient case is unique because of:

1. His early age at presentation (age 31);
2. The echocardiography results that showed only mild hypokinetic changes of the apex with normal cardiac BNP, which can mask the condition and delay proper diagnosis;
3. The early referral for CMR that led to the early detection of the potentially lethal LVNC.

CMR diagnostic criteria for LVNC use both long- and short-axis views across several segments. An end-diastolic ratio between non-compacted and compacted layers greater than 2.3 is considered diagnostic of myocardial non-compaction [5]. CMR provides highly sensitive (86%) and specific (99%) diagnostic value for LVNC, and may help with early diagnosis and intervention [5]. Early reports of mortality in adults with LVNC are approximately 50% within 6 years of diagnosis [1]. More recently, the observed mortality ranged from 4% to 12.7% over a period of 17 months to 4.5 years [9, 10]. Predictors of outcome include the development of ventricular arrhythmias, heart failure, and thromboembolism. Given the poor prognosis associated with LVNC, early diagnosis through CMR and timely intervention may change the course of the disease and prevent the development of complications.

CONCLUSION

While echocardiography is the primary modality for the diagnosis of LVNC, it is important to take a comprehensive approach to patient evaluation and not rely on a single diagnostic study or parameter. Furthermore, a routine use of CMR in the evaluation of frequent ventricular ectopic beats in symptomatic and otherwise asymptomatic patients was recently supported by the 2019 European Heart Rhythm Association (EHRA) Focus Guideline for the Management of Asymptomatic or Symptomatic Arrhythmia [11]. This should encourage physicians to lower the threshold for CMR use in this patient population.

ETHICS APPROVAL AND CONSENT TO PARTICIPATE

This study has been approved by the Ethics Committee of Emirates Specialty Hospital, Dubai Healthcare City, Dubai, UAE.

HUMAN AND ANIMAL RIGHTS

Not applicable.

CONSENT FOR PUBLICATION

Not applicable.

STANDARD FOR REPORTING

CARE guidelines have been followed in this case report.

FUNDING

None.

CONFLICTS OF INTEREST

The authors declare no conflict of interest, financial or otherwise.

ACKNOWLEDGEMENTS

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Table 1: contd....

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<tbody>
<tr>
<td>26/01/2019 to 08/08/2019</td>
<td>Regular patient follow-up (every 2 to 4 weeks). Gradual titration of anti-heart failure treatment. Significant improvement in palpitations, shortness of breath, and asthma symptoms.</td>
<td>Echocardiogram (14/04/2019) shows the resolution of previous apical LV hypokinesia.</td>
<td>Continue ramipril 10 mg PO od and bisoprolol 10 mg PO od. Follow-up visit scheduled for 3 months.</td>
</tr>
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</table>

ECG - electrocardiogram, CBC - complete blood count, TSH - thyroid stimulating hormone, LVH - left ventricular hypertrophy, CMR - cardiac magnetic resonance, PO - by mouth, od - once daily.
REFERENCES


