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Case Report
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Left Ventricular Non-compaction: A Family of Eleven Children

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Abstract

Left Ventricular Non Compaction (LVNC) is a rare congenital cardiomyopathy which can present at any age. Here in we present a case of an eleven old female who was admitted with complaints of shortness of breath since two years and echocardiography revealing low ejection fraction and LVNC. On detailed workup, it was found that LVNC was running in her family with many of her siblings having similar condition. Although genetic association of LVNC has been reported before, yet such strong family history has not been reported so far.

Keywords: Left ventricular non-compaction; Cardiomyopathy; Gene; Heart block

Introduction

Left Ventricular Non Compaction (LVNC) is a rare congenital cardiomyopathy that is characterised by thin compacted epicardial layer and extensive non-compacted endocardial layer with prominent trabeculations and deep recesses [1]. It may vary from mild to severe forms and may present at any age. It has a strong hereditary and genetic predisposition and variable penetrance as is well illustrated in present case.

Case Presentation

An eleven year old female child, resident of Middle East, born out of first degree consanguineous marriage presented with complain of shortness of breath since 2 years. It was gradually progressive, more on lying down position, reduced by rest associated with swelling of feet. On general physical examination, child was tachycardic (112 per minute), tachypneic (respiratory rate-36 per minute), pitting pedal oedema extending up to ankle, raised jugular venous pressure. Systemic examination revealed a displaced apex in 6th intercostal space lateral to mid-clavicular line with parasternal heave and ejection systolic murmur in fourth intercostal space left to the sternum. There were also bilateral basal crackles in chest. Echocardiogram revealed Left Ventricle (LV) dysfunction (LV ejection fraction (EF) – 20%) with LV non-compaction. There was no aortic stenosis, coarctation and large vessels had normal course.

On taking detailed history, it was found that in the family of 11 siblings; 8 of them have various degrees of LVNC. Six were females; out of six females two were normal; other four had mild apical LVNC. Youngest kid is 2 year old and he is having LVNC with left ventricular dysfunction (LVEF=35%) and eldest is 22 year old (Normal). Out of five males, one died at 15 years of age, he was suffering from dilated cardiomyopathy with CHF due to severe LVNC. Second male sibling has LVNC with LVEF=40% and LV is dilated (LVIDd =6.5 cm,). Third male sibling had LVNC with second degree heart block (Heart rate =80-100/minute). The fourth male sibling was normal, the youngest one is two years old also has apical LVNC with LVEF of 35%. There was no evidence of clot on echocardiography in any of the siblings. There was no history of cerebrovascular accident or syncope in any of the family members. Thus, suggesting a strong hereditary association of LVNC. The pedigree chart of the family shown in Figure 1.

The child was managed symptomatically with loop diuretics, beta blockers and oxygen support. Her condition improved in due course of days and was discharged.

Discussion

LVNC also known as “spongy myocardium” is a rare congenital condition which can present at any age. Its prevalence in general population is 0.014 to 1.3 percent [2].

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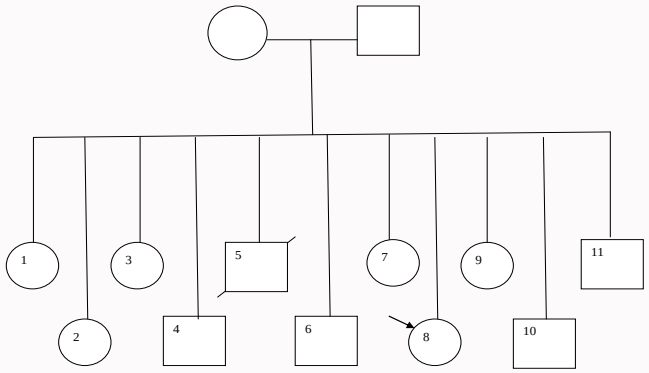

Figure 1: Pedigree of the family.
1) 22 years, Normal
2) 21 years, mild Apical LVNC
3) 20 years, Normal
4) 18 years, dilated LV, LVEF=45%
5) Died at 15 years
6) 13 years, mild Apical LVNC
7) 12 years, mild Apical LVNC
8) 11 years, LVNC, LVEF=20% (INDEX CASE)
9) 10 years, mild Apical LVNC
10) 2 years, mild Apical LVNC
11) 2 years, LVNC, LVEF=35%

Familial occurrence has been identified in 44% of reported cases [3]. Tafazzin gene located on chromosome Xq28 , expressed at high levels in cardiac and skeletal muscle has been implicated in LVNC pathogenesis in multiple studies [4-6]. Other genes including a-dystrobrevin, lamin A/C gene have been identified to have association with LVNC [3,7]. Post-mortem studies have discovered ischemic sub-endocardial lesions; thus supports the hypothesis that coronary microcirculation abnormalities may play a key role in its pathophysiology leading to contractile dysfunction. Diastolic dysfunction may be related to abnormal relaxation and restrictive filling caused by numerous trabecula. High percentage of associated neuromuscular disorders has been described in patients with LVNC [8].

In our case, one of the siblings presented with second degree heart block which is a rare presentation in this condition. However, Atrio-Ventricular (AV) block is a relatively common finding in pediatric series and is rarely reported in adult patients. The true cause of ventricular conduction abnormalities is still unclear. One of proposed mechanism is the fibrotic change in the conduction system develops gradually which may account for the progressive nature of the Atrio-ventricular block seen in adult patients [9]. A study by Xi Y et al. [10] recently showed that loss of function of hNav1.5 by a ZASP1 mutation is associated with intra ventricular conduction disturbances in left ventricular non-compaction.

Conclusion

The present case scenario suggests that LVNC has immense hereditary predilection and is expressed in both male and female phenotypes. However LVNC can present in various forms of severity varying from mild apical LVNC to severe DCM resulting in death. Since this disease is not present in every sibling, penetration of the culprit gene is not 100%.

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