

Maternal Death and Isolated Left Ventricular Non-Compaction Cardiomyopathy

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ABSTRACT

Left ventricular non-compaction cardiomyopathy (LVNC) is a rare congenital cardiomyopathy, which is characterized by hypertrabeculations and deep recesses of the left ventricle. A patient could be asymptomatic or presented with common manifestations, including reduced systolic function, arrhythmia, thromboembolic events and heart failure. The rarity of the condition as well as lack of proper assessment has probably led to this condition to be largely underdiagnosed or unrecognized. A 23-year-old lady had collapsed at home thirty one days after delivering her first child. She had a history of goitre diagnosed a year ago and noted to be fairly well throughout the pregnancy. Post mortem findings showed increased trabeculations of the left ventricle. Further history was obtained after the procedure, revealing symptoms such as syncopal attacks and bilateral lower limb weakness dated back as far as five years prior to her sudden demise. These features were in keeping with hypotension hypoperfusion effects resulted from reduced systolic function and decreased ejection fraction, as a result of left ventricular dysfunction. While LVNC remains a rare type of disease, we would like to highlight the importance of a good anamnesis. It may help to uncover some uncommon pathology such as this heart disease, thus warranting an appropriate cardiac imaging to be engaged to clinch the primary diagnosis.

Keywords: Primary cardiomyopathy; Maternal death; Non-compaction; Autopsy; Arrhythmia

INTRODUCTION

Left ventricular non-compaction cardiomyopathy (LVNC) is a rare type of cardiomyopathy, which resulted from an arrest of compaction during fifth to eighth week of embryogenesis.^{1,2} It is characterized by prominent trabeculations in the left ventricle and deep intertrabecular recesses which communicate with the ventricular cavity.^{3,4} Isolated LVNC is described when ventricular hypertrabeculation is present in the absence of any other co-existing congenital cardiac anomaly. World Health Organization (WHO) has initially categorized the disease as unclassified cardiomyopathy, and later re-categorized it as a genetic

cardiomyopathy.⁵ We present a case of isolated LVNC in a 23-year-old lady who appeared well and passed away approximately thirty one days after giving birth. The congenital defect was discovered at autopsy and further history obtained after the procedure revealed symptoms of the illness which had actually started a few years before her sudden demise.

CASE REPORT

A 23-year-old, gravida 1 para 1 lady was noted to be unresponsive at home and immediately brought to the nearest hospital. She was pronounced dead upon arrival and inquiry into a maternal death involved a post mortem examination. The history provided by the husband prior to the autopsy was brief; she had given birth thirty one days ago following an uncomplicated pregnancy and spontaneous vaginal delivery. She was diagnosed with hyperthyroidism approximately one year before the pregnancy and was under combined clinics follow-up throughout the pregnancy. The ultrasound imaging of the neck showed diffuse goitre. She was prescribed with Carbimazole and claimed to be compliant with medications. She was also noted to be having anaemia in pregnancy. After giving birth, her thyroid function tests were reviewed and she was discharged well. The last reading showed thyroid

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stimulating hormone (TSH) level was 0.001 nU/ml and the free T4 level was 17.55 nU/ml. Antithyroid globulin antibody was positive.

Autopsy examination showed a thinly built adult female measuring 152 cm in length and 51 kg in weight. Bluish discoloration was noted on the nail beds, indicating cyanosis. The anterior aspect of the neck was symmetrically enlarged. There was no significant injury noted in the body. Internal examination showed bilaterally enlarged thyroid glands. The right and left glands weighed 65 gm and 55 gm respectively. Cut surfaces of the glands revealed homogenous parenchyma. There was no area of haemorrhage or necrosis noted. The heart weighed 310 gm. In view of her body weight, a normal heart would weigh between 204 gm to 214 gm, thus indicating a cardiomegaly. Biventricular hypertrophy was noted and the right and left ventricular chambers were dilated. Upon sectioning, deep trabeculations were noted to affect approximately one third of the left ventricular wall thickness and most pronounced at the apex (Fig. 1). The coronary arteries were patent. Other internal organs appeared grossly normal. The uterine involution was also normal.

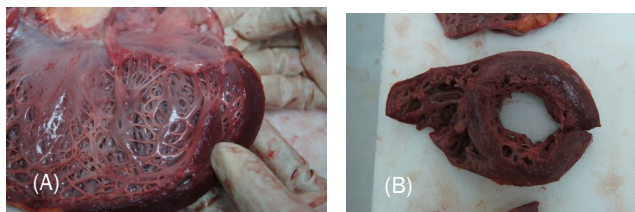


Fig. 1. (A) Cut section of the left ventricle showing subendocardial fibrosis and fine trabeculations. (B) Deep recesses and increased trabeculations are seen, consistent with LVNC.

Histopathology examination showed patchy fibrosis and hypertrophic cardiomyocytes, denoting past ischaemic events. The thyroid glands showed infiltration by mature lymphocytes and Hürthle cell metaplasia. These features were consistent with Hashimoto's thyroiditis. Blood for alcohol and toxicology screening were also obtained and the results were negative for common drugs.

The cause of death was given as left ventricular non-compaction cardiomyopathy. The possible mechanism of death was cardiac arrhythmia.

DISCUSSION

LVNC is being increasingly recognized and diagnosed with the advancement of echocardiography and cardiac MRI.⁴ At present, there is no consensus on the diagnostic criteria for the condition; however, some authors rely on the thickness of the trabeculated mass of more than 20% of the total mass.⁶ The increased non-compacted layer of the ventricular wall gives the appearance of a spongy subendocardium.⁶ Several genes have been reported

to be associated with this condition, including mutations of Z-band alternatively spliced PDZ-motif protein (ZASP), α -dystrobrevin (DTNA), tafazzin (TAZ-G4.5) and genes encoding sarcomeric proteins.⁴

This case presented with brief medical history obtained from the husband. Apart from the goitre, she was said to be healthy. At autopsy, the feature of increased trabeculations of the left ventricle was not convincing enough to the pathologist in order to write off the cause of death to be due to LVNC. Hence, further medical history was sought from the next-of-kin. To our surprise, stories unfolded interestingly. According to the mother, the deceased was healthy throughout her childhood. After finishing school approximately five years ago, she started to complain of difficulty in breathing following heavy physical exertion. She also began to develop episodes of syncopal attack and numbness of the lower limbs. Our hospital record showed that she actually presented here approximately three years ago due to bilateral lower limb weakness. Previous episodes actually began a year before her first admission to this hospital. The diagnosis made was hypokalemic periodic paralysis due to possible electrolyte imbalance and she was discharged without further follow up or investigation. For the past two years also, she had at least five syncopal attacks associated with lower limb weakness which lasted for several hours. She also complained of lethargy and became less and less active. Somehow, this important piece of information was not communicated to the attending physicians at the combined clinics during her ante-natal visits as well as the post-natal follow-ups. After giving birth, her condition slightly worsened and she was less mobile for the past few days prior to death.

Clinical manifestations in LVNC ranging from completely asymptomatic to end-stage heart failure.⁷ Symptoms are generally associated with left ventricular dysfunction, hence causing reduced systolic function, thromboembolic event, arrhythmia and eventually heart failure.^{1,6,8} Sudden cardiac death may occur as a result of ventricular tachyarrhythmia.⁷

Ultimately, the clinical symptoms fit the cardiac condition, we thought she had. The syncopal attacks, the lower limb weakness and the lethargy are most probably associated with hypotension hypoperfusion effects resulted from reduced systolic function and decreased ejection fraction. Biventricular involvements in this case as well as her deteriorating conditions after giving birth are probably an indicator that the impaired left ventricular function is progressing towards heart failure.

LVNC usually has no specific findings on histology examination. Interstitial fibrosis and endocardial fibroelastosis may be found in some cases.⁴ In this case, the left ventricle demonstrates patchy

fibrosis, in keeping with episodes of ischaemic events in the past and a possible mechanism for arrhythmia.

We have considered peripartum cardiomyopathy due to Hashimoto's thyroiditis and Tako-Tsubo cardiomyopathy (TTC) in this patient. Hashimoto's thyroiditis can cause cardiomegaly and left ventricular dysfunction and global hypokinesia associated with pericardial effusion.⁹ This patient did not have pericardial effusion. The clinical presentation in TTC can be dramatic including acute heart failure or cardiogenic shock due to severe impairment of left ventricular function. It is generally due to rapid increase of thyroid hormones either iatrogenic or endogenous. The mechanisms by which hyperthyroidism trigger TTC remains poorly understood. In the presence of dysthyroid states the rapid increase in thyroid hormones might result in acute activation of adrenergic system which plays a major role in the pathogenesis of TTC. The histological evidence; although it shows evidence of past ischaemic events, does not support the diagnosis of TTC.¹⁰

Further reviews of isolated LVNC showed that 35% of patients diagnosed with LVNC actually presented with normal left ventricular size and thickness.⁷ In other instances, the diagnosis was made in the absence of any other underlying congenital cardiac anomalies.¹ In concluding this case, the pathologist arrived to the diagnosis of LVNC based on the gross autopsy findings in combination with the previous complaints of non-specific symptoms. There was no other cardiac abnormality or disease pathology observed.

As a conclusion, LVNC is still widely underdiagnosed. Good anamnesis may help to uncover some rare pathology such as this heart disease, thus warranting an appropriate cardiac imaging to be engaged and to clinch the primary diagnosis.

Conflict of interest

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Ethical approval

None.

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