A Case Report of Infant with Left Ventricular Noncompaction

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Left ventricular noncompaction (LVNC) is a rare cardiomyopathy resulting from abnormal arrest during endomyocardial embryogenesis. The authors present a 6-month-old infant with intractable cardiogenic shock and echocardiographic features of LVNC, characterized by excessively prominent ventricular trabeculation and deep intertrabecular recesses as the first case report in Thailand.

Keywords: *Left ventricular noncompaction, Cardiomyopathy, Heart failure, Arrhythmia, Embolization*

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Left ventricular noncompaction (LVNC) is a rare cardiomyopathy characterized by excessively prominent ventricular trabeculation and deep intertrabecular recesses, resulting from abnormal arrest during endomyocardial embryogenesis[1]. LVNC has been initially described in the pediatric population, however recent studies also reported in adults[1,2]. The authors described an infant who presented with clinical and echocardiographic features of LVNC, the first reported case in Thailand.

Case Report

A 6-month-old Thai male infant was referred due to shock. His mother noticed a right upper quadrant mass about 1 month prior to seeking treatment without other symptoms followed 3 weeks later by edema and poor feeding. At admission to the hospital, he had profound shock, a thread femoral pulse with a rate of 150/min, blood pressure of 62/50 mmHg, respiratory rate of 60/min, and room air cutaneous oxygen saturation of 85%. This patient was afebrile, body weight of 4,600 grams and length of 61.3 cm. Physically he had low set ears without other dysmorphic features. The cardiac auscultation demonstrated poor heart tone with normal S_1, S_2, and no definite murmur. There was no adventitious breath sound. The liver span was 10 cm and 5 cm below right costal margin with firm consistency, and no splenomegaly.

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He was the second child in the family with the birth weight of 3,000 grams. His past medical history was uneventful, however the developmental milestone was globally delayed especially in gross motor area. There was no history of consanguinity, sudden cardiac death or early infant death in family members.

The chest X-ray revealed pulmonary venous congestion and cardiomegaly with the cardiothoracic ratio of 0.62. The electrocardiogram showed sinus rhythm with the rate of 150/min, P axis 15°, QRS axis 45°, normal PR and QRS interval, no definite chamber enlargement, and ST depression with T-wave inversion in V_{3-5} (Fig. 1). The echocardiography demonstrated remarkable reduction of the left ventricular systolic function (ejection fraction 10%) and prominent trabeculations with deep intertrabecular recesses, predominately in the apical region without other structural cardiac defects (Fig. 2). The complete blood count showed hemoglobin 10.3 gm/dl, hematocrit 35%, total white blood cell count 14,700 cell/mm^3, neutrophil 34%, lymphocyte 59%, platelet 314,000 cell/mm^3. The serum urea nitrogen 11.4 mg%, creatinine 0.8 mg%, sodium 134 mmol/l, potassium 8.0 mmol/l, chloride 102 mmol/l, total CO_2 8 mmol/l, and blood sugar 119 mg%. The liver function test revealed total bilirubin 2.40 mg%, direct bilirubin 2.38 mg%, AST 229 U/L, ALT 136 U/L, alkaline phosphatase 386 U/L, total protein 5.1 gm%, and albumin 2.8 gm%. The non-fasting cholesterol 216 mg%, triglyceride 1,059 mg%, HDL-C 13.7 mg%, total LDH 2,203 U/L, total CPK 452 U/L, and CK-MB 12.4 ng/ml. The arterial blood gas

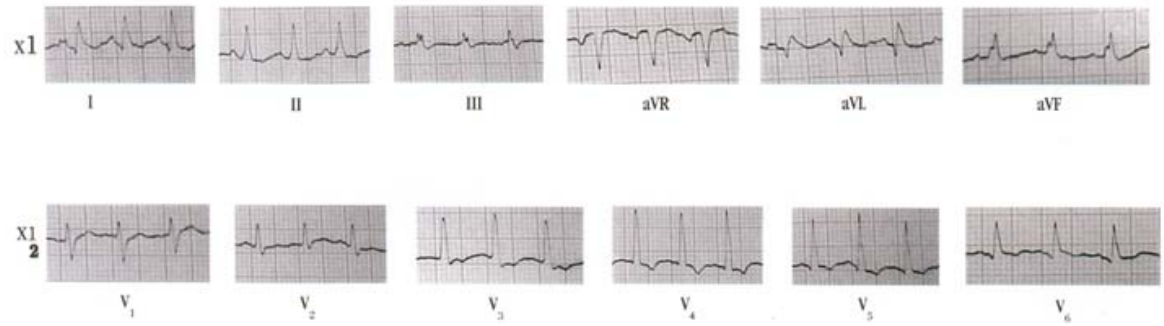

Fig. 1 Electrocardiogram demonstrates ST depression in V_3 and T-wave inversion in V_{3-5}

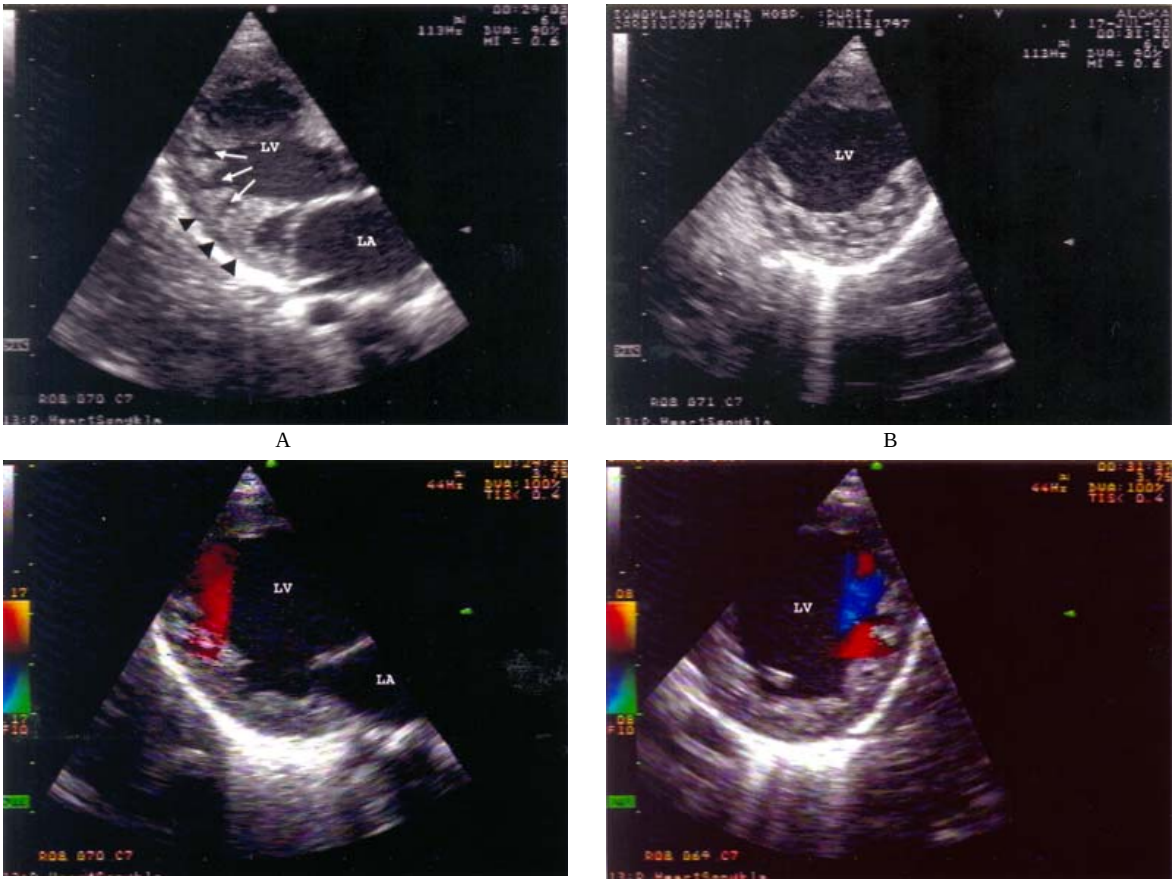

Fig. 2 Echocardiogram of patient, (A) Parasternal long- axis view, there is a thin epicardial layer (arrowheads) and an extremely thickened endocardial layer with numerous prominent left ventricular trabeculations, increased depth of intertrabecular recesses (thin arrow). (B) The 2 layers are best visualized at end systole as shown in this short axis view. (C) There is blood flow from the ventricular cavity into the deep recesses visualized on color Doppler study. (D) Shows typical forward (red color) and reversed flow (blue color) between prominent trabeculations during the cardiac cycle. LA = left atrium, LV = left ventricle

demonstrated pH 7.04, pCO_2 14.4, pO_2 57.9 mmHg and base excess -25 mmol/L.

Dopamine and dobutamine were initially infused 10 mcg/kg/min each. Thiamine was also administered for therapeutic trial of cardiac beriberi. The metabolic acidosis persisted though repetitive doses of bicarbonate were given. Dopamine and dobutamine were maximized with the rate of 20 mcg/

kg/min with additional of adrenaline infusion 2 mcg/kg/min, without response to cardiogenic shock. Five hours after admission, he developed monomorphic ventricular tachycardia, which resisted all medical modalities. The infant eventually died from malignant ventricular fibrillation. A cardiac and liver necropsy was performed post mortem, however cardiac muscle tissue was not adequate and it showed generalized fatty change of liver parenchyma. Autopsy was unable to perform due to religious belief. All family members were completely examined for cardiovascular diseases including echocardiography, and all of the investigations were normal.

Discussion

LVNC is an unclassified cardiomyopathy according to the WHO classification which has a variety of spectrum ranging from asymptomatic to a fulminant of left ventricular dysfunction, ventricular arrthymia, systemic embolization, even sudden death syndrome[2,3]. Although an abnormality EKG of LVNC is non-specific but mostly abnormal and may be malignant arrthymia[1-4]. In the presented patient, the EKG also showed ST-T segment and T wave abnormalities and fatal ventricular arrhythmia. Although many modalities such as computer tomography and magnetic resonance imaging have been developed to detect more subtle variants of non-compaction, however no diagnostic criteria have yet been proposed. Currently, the hallmark of the diagnosis still base on the echocardiographic criteria. The presented patient came with severe left ventricular failure and malignant ventricular arrthymia; he fulfilled 4 echocardiographic criteria of LVNC described by Jenni et al[5], which are the most widely known of valid echocardiographic criteria by agreement with the necropsy finding, as follows: 1) No coexisting cardiac abnormalities. 2) A two layer structure was seen, with a compacted thin epicardial band and a much thicker noncompacted endocardial layer of trabecular meshwork with deep intertrabecular recesses. A maximal end systolic ratio of noncompacted to compacted layers of 2.2. 3) Predominant area was in the apical and mid-ventricular area. 4) There was Color Doppler evidence of deep perfused intertrabecular recesses. The hypertriglyceridemia and fatty liver in the presented patient has not yet been described in LVNC. The authors postulated that it may result from his severe protein caloric malnutrition; nevertheless, fasting triglycerides > 1,000 mg/dl in children reflect severe hyperchylomicronemia, indicate an underlying genetic disorder, and increase risk of premature atherosclerosis[6]. However, it too early onset for myocardial infarction and result of lipid profile in his first-degree relatives were unremarkable so we still could not make a definite conclusion for his hyperlipidemia. Both familial and sporadic forms of LVNC have been described. Familial form has been observed in 19-44% in the pediatric population[3,7] and about 18% in adult[2]; however, the presented patient was a sporadic case. Gene G4.5 on the Xq28 chromosomal region is responsible for some familial cases, especially in X-link cases; those for the sporadic forms of LVNC have not yet been described.[8] Facial dysmorphisms described by Chin et al was low-set ears, prominent forehead, strabismus, high arched palate, micrognathia and motor delay[1]. Some cases of LVNC have been found to be associated with certain genetic syndrome as follows; Barth syndrome, Melnick-Needles, mutations in the gene encoding α-dystrobrevin and Cypher/ZASP, and distal 5q deletion[8,9]. The presented patient had only low-set ears and motor delay, which are unable to conclude a definite genetic syndrome. The major limitations of this report are the following: a rapid deterioration of patient during admission, retrospective review of the literatures, and limitation of some investigations at our center; therefore, genetic study, skeletal biopsy, neurological investigation, and metabolic screening were unable to obtain for more information.

The diagnosis of LVNC is mostly missed due to lack of recognition. The authors hope that the present report will lead to more awareness of LVNC, especially in cases of clinical presentation of left sided heart failure, ventricular arrthymia, systemic embolization or cardiomyopathy. Echocardiography is the best method to confirm the diagnosis. Further study such as lipid profiles, metabolic screening, and skeletal biopsy, and genetic study are necessary to elucidate its association with LVNC.

References

1. Chin TK, Perloff JK, Williams RG, Jue K, Mohrmann R. Isolated noncompaction of left ventricular myocardium: a study of eight cases. Circulation 1990; 82: 507-13.
2. Oechslin EN, Attenhofer Jost CH, Rojas JR, Kaufmann PA, Jenni R. Long-term follow-up of 34 adults with isolated left ventricular noncompaction: A distinct cardiomyo-pathy with poor prognosis. J Am Coll Cardiol 2000; 36: 493-500.
3. Ichida F, Hamamichi Y, Miyawaki T, Ono Y, Kamiya T, Akagi T, et al. Clinical features of isolated non

compaction of the ventricular myocardium: long-term clinical course, hemodynamic properties, and genetic background. J Am Coll Cardiol 1999; 34: 233-40.
4. Yasukawa K. Terai M, Honda A, Kohno Y. Isolated Noncompaction of Ventricular Myocardium Associated with Fatal Ventricular Fibrillation. Pediatr Cardiol 2001; 22: 512-4.
5. Jenni R, Oechslin E, Schneider J, Jost CA, Kaufmann PA. Echocardiographic and pathoanatomical characteristics of isolated left ventricular non-compaction: a step towards classification as a distinct cardiomyopathy. Heart 2001; 86: 666-71.
6. Andrew M, Tershakovec and Daniel JD. Disorders of lipoprotein metabolism and transport. In: Behrman RE, Kliegman RM, Jenson HB, editors. Nelson textbook of pediatrics.17th ed. Philadelphia: WB Saunders, 2004: 445-59.
7. Pignatelli RH, McMahon CJ, Dreyer WJ, Denfield SW, Price J, Belmont JW, et al. Clinical characterization of left ventricular noncompaction in Children: A relatively common form of cardiomyo-pathy. Circulation 2003; 108: 2672-8.
8. Zambrano E, Marshalko SJ, Jaffe CC, Hui P. Isolated noncompaction of the ventricular myocardium: clinical and molecular aspects of a rare cardiomyopathy. Laboratory Investigation 2002; 82: 117-22.
9. Murphy RT, Thaman R, Blanes JG, Ward D, Sevdalis E, Papra E, et al. Natural history and familial characteristics of isolated left ventricular non-compaction. Eur Heart J 2005; 26: 187-92.

รายงานทารกที่มีความผิดปกติของกล้ามเนื้อหัวใจห้องล่างซ้ายชนิด noncompaction

สุภาพร โรยมณี, วรการ พรหมพันธุ์, สมเกียรติ โสภณธรรมรักษ์

รายงานทารกเพศชายอายุ 6 เดือนที่มีความผิดปกติของกล้ามเนื้อหัวใจห้องล่างซ้ายชนิด noncompaction ซึ่งเป็นภาวะที่พบได้น้อยมาก ผู้ป่วยมีอาการหัวใจวายที่ไม่ตอบสนองต่อการรักษา ผลการตรวจคลื่นเสียงสะท้อนหัวใจพบความผิดปกติของ trabeculation และการบีบตัวของหัวใจห้องล่างซ้าย เป็นรายงานแรกของประเทศไทยสำหรับผู้ป่วยที่มีภาวะนี้