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Siblings with left ventricular diverticulum and hypertrophic cardiomyopathy

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Summary

This report describes very rare siblings who had a left ventricular muscular diverticulum and hypertrophic cardiomyopathy.

The first case is a 16-year-old male. On two-dimensional echocardiography, a left ventricular muscular diverticulum in the posterior wall and mitral valve prolapse were detected. The former was verified by left ventriculography. Endomyocardial biopsy showed findings compatible with hypertrophic cardiomyopathy.

The second case is a 13-year-old female. Two-dimensional echocardiography revealed a left ventricular muscular diverticulum in the same location as that of the first case, and mitral valve prolapse as well. The former was confirmed by left ventriculography. The endomyocardial biopsy findings were compatible with hypertrophic cardiomyopathy. The coronary angiograms were normal in both cases.

To our knowledge, familial appearance of a left ventricular diverticulum has not yet been reported, and a left ventricular diverticulum at the posterior wall in cases with hypertrophic cardiomyopathy is very rare.

Key words

Left ventricular diverticulum　Hypertrophic cardiomyopathy　Siblings　Familial appearance
Two-dimensional echocardiography　Left ventriculography

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Introduction

Left ventricular diverticulum is a rare congenital anomaly. To the present time, two types of left ventricular diverticulum have been reported[1] ; one is a muscular type, the other, a fibrous type. Reported here are siblings, a brother and younger sister, each with a left ventricular diverticulum associated with hypertrophic cardiomyopathy (HCM). These left ventricular diverticula were located in the posterior walls of the left ventricles. They may be the first such diverticula to be reported in siblings having associated HCM.

Case reports

Case 1

When this 17-year-old male was 15 years of age, an ECG abnormality was noted during a routine examination at his high school. He experienced two episodes of syncope during exercise, each lasting for several minutes, and was admitted to our department. His admission blood pressure was 112/64 mmHg ; his pulse rate was 60/min and regular. On auscultation, the second heart sound was widely split during the inspiratory phase. A grade 3/6 ejection systolic murmur was audible in the third left intercostal space near the sternum and at the apex. The liver edge was palpable four fingerbreadths below the RCM in the epigastrium. Normal sinus rhythm and ST segment depression were observed in leads I, II, III, aVF, and $V_3 \sim V_6$ in the electrocardiogram. T waves were inverted in the II, III, and aVF leads. High voltage was observed in the left lateral precordial leads, and left atrial overload, as well (**Fig. 1A**). His chest radiograph showed abnormal prominence of the right heart border (**Fig. 2**). Two-dimensional echocardiography revealed left ventricular wall hypertrophy and a left ventricular muscular diverticulum in the posterior wall of the left ventricle, which contracted during the systolic phase. Mitral valve prolapse was also observed, and the left atrium was enlarged (**Fig. 3A and B**). An increased A/R ratio of left ventricular filling flow was shown by pulsed Doppler echocardiography. The resting hemodynamic data

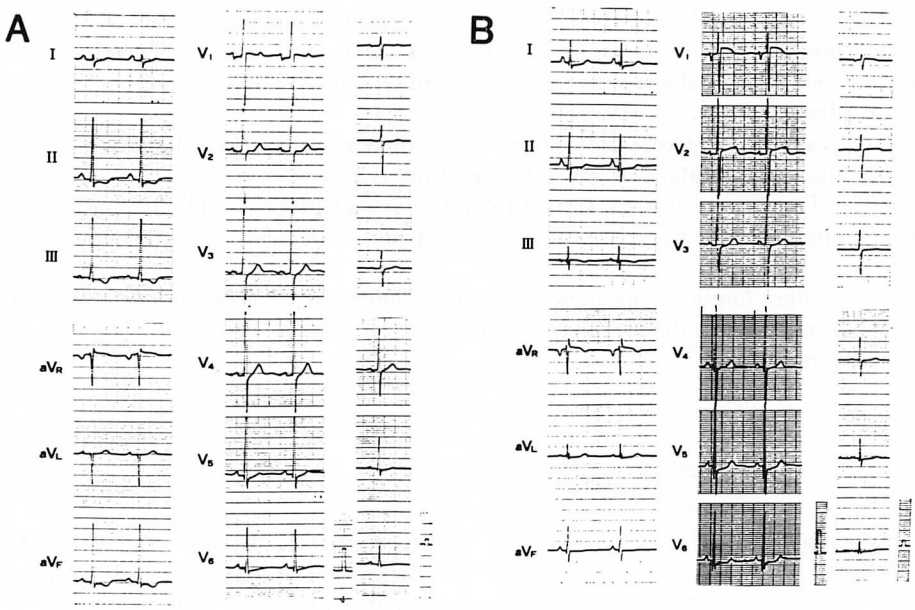

Fig. 1. Electrocardiograms of Cases 1 (A) **and 2** (B).

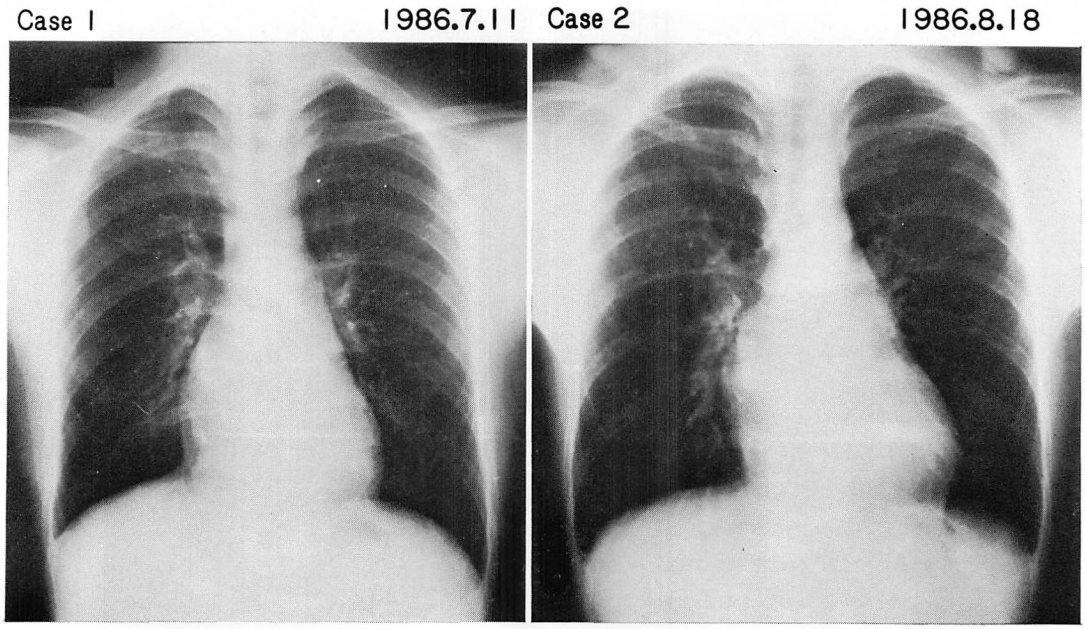

Fig. 2. Chest radiographs of Cases 1 and 2.
Case 1: the cardiothoracic ratio is 0.47. Case 2: slight cardiac enlargement is noted and the cardiothoracic ratio is 0.52.

of each chamber obtained by catheterization are shown in **Table 1**. The pulmonary capillary wedge pressure and left ventricular end-diastolic pressure were abnormally elevated. A left ventriculogram confirmed the presence of a left ventricular diverticulum in the posterior wall, which contracted during the systolic phase. In addition, a mitral regurgitant murmur of grade II was detected (**Fig. 4A and B**). Coronary angiogram was normal. A variety of studies ruled out secondary hypertrophic cardiomyopathy. Results of endomyocardial biopsy included hypertrophic myocytes, myocardial degeneration, interstitial fibrosis and bizarre myocardial hypertrophy with disarray (**Fig. 5a**). These findings are compatible with HCM. Intravenous pyelography revealed double ureters on the left side.

Case 2

This 13-year-old patient was the sister of Case 1. On physical examination, her blood pressure was 84/42 mmHg; pulse rate, 68/min and regular. Auscultation revealed the second heart sound to be widely split in the inspiratory phase. A grade 3/6 ejection systolic murmur was audible in the third left intercostal space near the sternum. On electrocardiography, ST segment depression was observed in leads I, II, aV_L, and $V_3 \sim V_6$, and ST segment elevation in leads $V_1 \sim V_2$. The T waves were inverted in lead III, and isoelectric in lead aV_F. High voltage was also seen in the left lateral precordial leads, and left atrial overload as well (**Fig. 1B**). His chest radiography revealed prominence of right and left heart borders (**Fig. 2**). Two-dimensional echocardiography showed a left ventricular muscular diverticulum in the posterior wall of the left ventricle, which also contracted during the systolic phase (**Fig. 3C and D**). The left atrium was enlarged; the basal portion of the interventricular septum was slightly hypertrophic; and mitral valve prolapse was observed. An increased A/R ratio of left ventricular filling flow was revealed by pulsed Doppler echocardiography. The cardiac

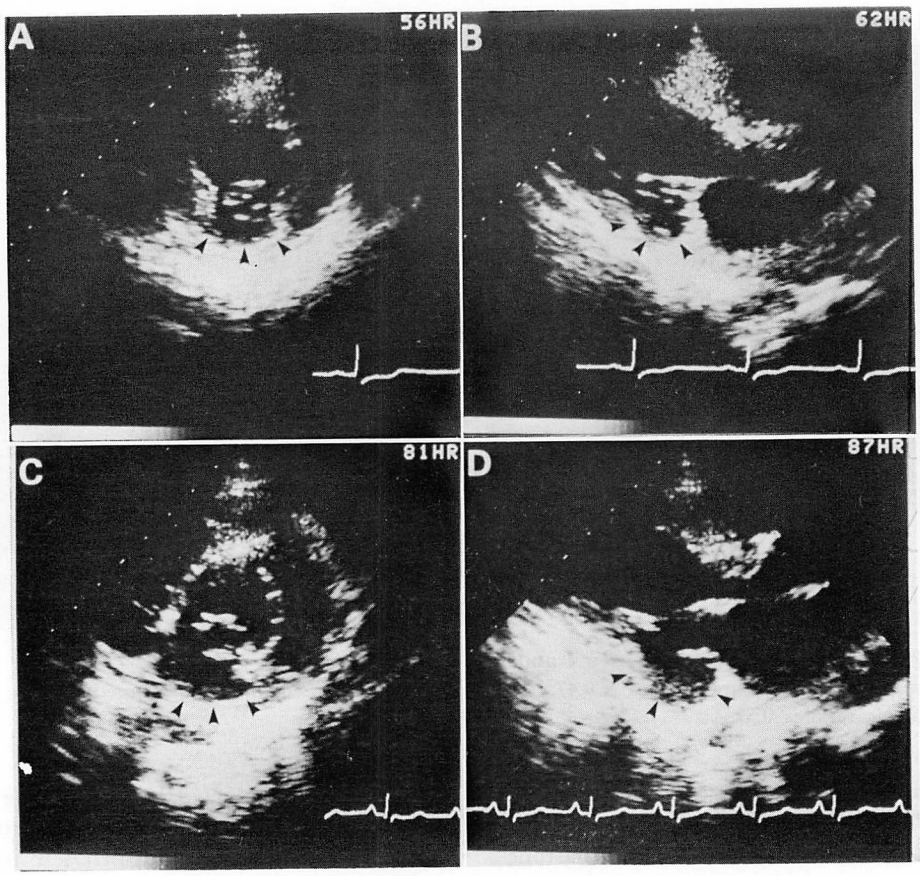

Fig. 3. Two-dimensional echocardiograms of the siblings.

Short-axis view (A) and long-axis view (B) of Case 1 in end-diastole. Short-axis view (C) and long-axis view (D) of Case 2 in end-diastole.

Arrowheads indicate a left ventricular diverticulum.

Table 1. Hemodynamic data of the two cases

	Case 1	Case 2
Right atrium (a/v/mean, mmHg)	8/ 3/ 3	6/ 4/ 4
Right ventricle (s/d/ed, mmHg)	43/ 1/ 6	33/ 0/ 5
Pulmonary artery (s/d/mean, mmHg)	33/14/23	32/14/21
PCWP (a/v/mean, mmHg)	19/26/18	14/21/12
Left ventricle (s/d/ed, mmHg)	100/10/34	90/ 6/24
Aorta (s/d/mean, mmHg)	102/70/88	114/71/89
Cardiac index (l/min/m^2)	3.1	2.9

a=a wave; v=v wave; s=systole; d=diastole; ed=end-diastole; PCWP=pulmonary capillary wedge pressure.

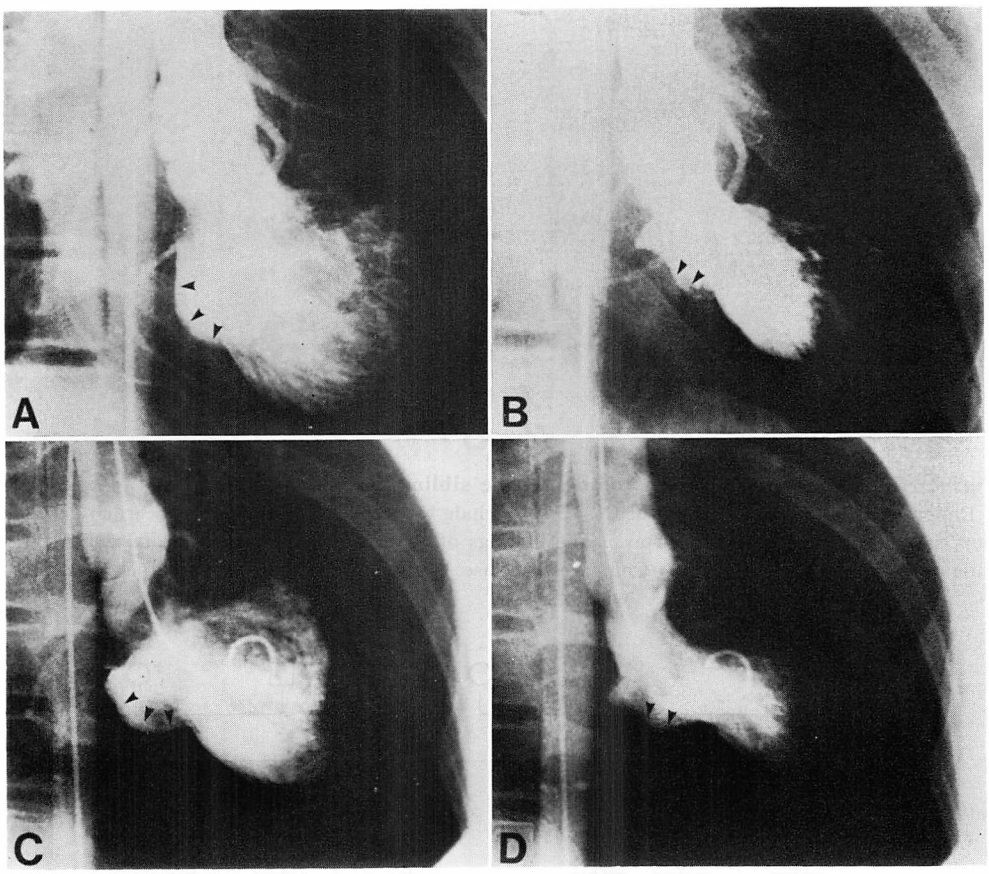

Fig. 4. Right anterior oblique view of ventriculograms of Cases 1 and 2 (C and D) **in end-diastole** (top) **and in end-systole** (bottom).
Arrowheads indicate a left ventricular diverticulum which contracts during systole.

catheterization data are shown in **Table 1**. As in Case 1, high pulmonary capillary wedge pressure and left ventricular end-diastolic pressure were observed. Left ventriculography confirmed the presence of a left ventricular diverticulum in the posterior wall (**Fig. 4C and D**). This diverticulum contracted synchronously in the systolic phase. The coronary angiogram was normal except for a shift of the branches of the right coronary artery caused by the left ventricular diverticulum. Results of endomyocardial biopsy included myocardial degeneration, relatively strong interstitial fibrosis and disarray. As in Case 1, the biopsy findings were compatible with HCM. However, interstitial fibrosis was milder than in Case 1 (**Fig. 5b**). According to the HLA typing study, AW 33, B 12, and DRW 11 in these two cases are common. Chromosomal studies were normal in both cases. Investigation of 18 relatives by ECG and chest radiography revealed no finding suspicious of HCM (**Fig. 6**).

Discussion

Left ventricular diverticulum is a rare congenital abnormality which has two types[1]. One is muscular[1~16]; and the other, fibrous[17~20]. Muscular diverticula are usually located at

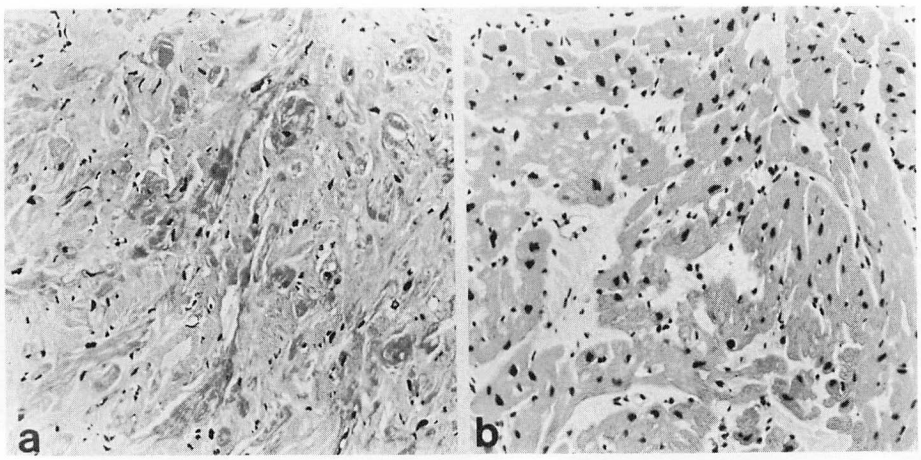

Fig. 5. Endomyocardial biopsy findings of the sibling cases.

Endomyocardial biopsy findings of Case 1 (a) include hypertrophic myocytes, myocardial degeneration, interstitial fibrosis and disarray. The findings of Case 2 (b) include myocardial degeneration, interstitial fibrosis and disarray. Compared to Case 1, interstitial fibrosis was mild in Case 2.

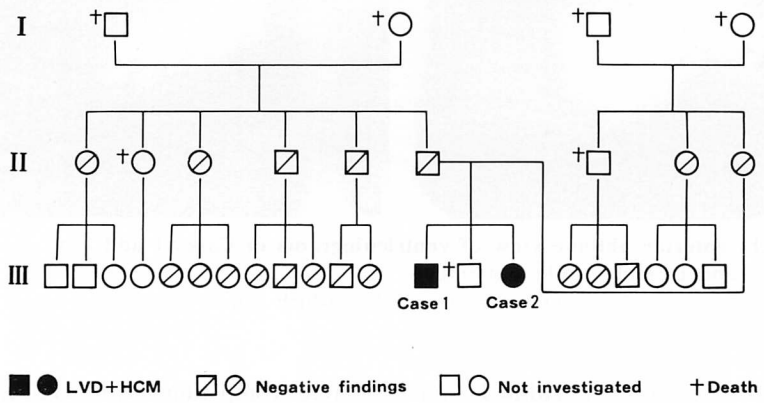

Fig. 6. Pedigree chart of the family.

Eighteen relatives had no abnormal findings suggestive of HCM by ECG and chest radiography.

LVD+HCM=left ventricular diverticulum associated with hypertrophic cardiomyopathy; Negative findings=no abnormal findings suggestive of HCM by ECG and chest radiography; Not investigated=relatives not investigated by ECG and chest radiography.

the apex or the inferoposterior wall. The wall of the muscular diverticulum consists of intact muscle layers or muscle fibers and some fibrous tissue[1]. Furthermore, many cases of left ventricular muscular diverticulum arise from the apex and are complicated by midline defects (abnormalities of the abdominal wall, sternum, and diaphragm)[1,8~10]. However, relatively small left ventricular muscular diverticula without midline defects as opposed to left ventricular diverticula arising from the apex, have also been reported[2,11~15]. Left ventri-

cular muscular diverticula can be differentiated from fibrous ones by virtue of their contractility during the systolic phase. Fibrous diverticula are usually apical or subvalvular in position, and their walls are composed of fibrous tissue[17~20]. The diverticula in the present two cases, both contracted in the systolic phase according to left ventriculography and two-dimensional echocardiography. Both of these diverticula were therefore regarded muscular. Until now, six cases of left ventricular diverticulum with HCM have been reported[3~6]. Three of them are thought to have had muscular diverticula. However, the diverticula of all six cases were located at the apex, therefore different from our two cases, whose diverticula were in the posterior wall. Our cases may be the first reported left ventricular muscular diverticula in the posterior walls associated with HCM. Furthermore, our review of the literature showed that left ventricular diverticula in siblings have not yet been reported[16]. Therefore, our patients may be the first in which the familial appearance of left ventricular diverticulum was observed. Furthermore, the diverticula of both of our cases were in the posterior walls of the left ventricles and associated with HCM, and such a combination is extremely rare. The HLA typing in our cases is usually observed in the familial type of HCM in Japan[21]. No characteristic HLA typing has been demonstrated in this disorder, as in our cases. Though there was no clear evidence of genetic factors, we must consider the role of a genetic background in the association of left ventricular diverticulum and HCM in the present cases, because the left ventricular diverticula were in the same regions of the left ventricles.

要　約

特発性肥大型心筋症に合併した家族性左室憩室の2症例

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飯村　攻

肥大型心筋症（HCM）で左室後壁に限局した筋性左室憩室を合併した同胞例のまれな症例を経験したので報告した．第1例は16歳，男性，断層心エコー図上，左室後壁に限局した筋性左室憩室および僧帽弁逸脱を認めた．左室造影で筋性左室憩室を確認し，右室心筋生検により HCM が確診された．第2例は13歳，女性，断層心エコー図上，兄と同一部位の筋性左室憩室と僧帽弁逸脱を認めた．左室造影で同部位に筋性左室憩室を認め，右室心筋生検は，典型的 HCM の像を呈した．両例とも冠動脈に狭窄所見を認めなかった．

近年，断層心エコー図法の普及により，筋性左室憩室の報告が増加している．しかし，我々の調べ得た限りでは，家族内発症の報告はなく，本報告が初めての症例と思われた．HCM と心尖部筋性左室憩室の合併例の報告は3例みられるが，左室後壁に存在する例としては最初の症例と考えられる．

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