Compressed Heart and Mitral Valve Prolapse in a Case of Duchenne’s Progressive Muscular Dystrophy with Thorax Deformity

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SUMMARY

We report on a 15 year old patient with Duchenne’s progressive muscular dystrophy who demonstrated a narrowing of the left ventricular inflow and outflow tracts due to compression by a highly deformed thoracic spine. A systolic murmur (4/6) with thrill and a diastolic murmur (2/6) were heard, with these murmurs being louder in the expiratory phase as compared with the inspiratory phase. The second heart sound showed a paradoxical splitting. Echocardiograms revealed a compressed and narrowed left ventricle and a prolapsed mitral valve. The intensities of the heart murmurs changed synchronously with the chamber’s narrowing due to respiration. A narrowed left ventricle occurring as a result of the compression by the deformed thoracic spine is thought to be the cause of these cardiac findings.

Additional Indexing Words:
Thorax deformity Compressed left ventricle Mitral valve prolapse Stenotic heart murmurs Duchenne’s progressive muscular dystrophy

IN progressive muscular dystrophy, the thorax and thoracic spine become deformed as cardiopulmonary function decreases.1)-7) Recently, echocardiography has provided a satisfactory noninvasive method to evaluate the cardiac anatomy and cardiovascular hemodynamics.8),9) We report on a case with Duchenne’s progressive muscular dystrophy with high grade deformations of the thorax and thoracic spinal column who showed 1) a narrowed...
cavity of the left ventricle and probably subsequent stenotic murmurs of the outflow and inflow tracts of the left ventricle, and 2) mitral valve prolapse.

**Case Report**

The patient is a male, 15 years old, who has been hospitalized since the age of 6. His delivery was full-term and normal and he was in good health until the age of 3 when it was observed that he fell down while trying to walk. At that time, due to the findings of elevated CPK and LDH, and skeletal muscle biopsy, he was diagnosed as having Duchenne's progressive muscular dystrophy. Until recently, no doctor noted significant murmurs.

*Physical findings:* An examination disclosed a generalized muscle atrophy with deformities in the extremities and trunk. His thorax was deformed

![Fig. 1. Chest radiogram. High grade scoliosis and lordosis are shown.](image1)

![Fig. 2. Chest CT image. The heart is compressed between the sternum and the thoracic spine.](image2)
and flat, and the thoracic spine showed a high-degree of scoliosis and lordosis. An auscultation indicated the lungs were clear. The heart was not enlarged and had a normal first sound and a paradoxically split second heart sound. At the apex, a grade 2/6 diamond-shaped systolic murmur, comprised of both a rumbling and high-pitched diastolic murmur, was heard. The systolic murmur was loudest around the third and fourth intercostal space (ICS) of the left sternal border (grade 4/6 with systolic palpable thrill) and radiated toward the neck. Both the systolic and diastolic murmurs became louder during the expiratory phase as compared with the inspiratory phase.

Chest radiography showed a scoliotic and lordotic thoracic spine with a consequent flattened thorax (Fig. 1). A chest CT film at the heart level showed a flattened thorax with an anteriorly displaced thoracic spine (Fig. 2). Phonocardiograms showed systolic and diastolic murmurs at the fifth ICS on the left sternal border (Fig. 3-a). Respiratory changes with expiratory accentuation of both the systolic and diastolic murmurs were apparent (Fig. 3-b).

**Fig. 3.** Phonocardiograms. a) Systolic murmur (SM) and diastolic murmur (DM) are shown at 5th intercostal space of the left sternal border (5LSB). b) Both murmurs increased during expiration. Carot. P= carotid pulse.
Fig. 4. Echocardiograms. a) Two-dimensional echocardiograms serially connected. The left ventricular cavity is narrowed at the base by the compression of the thoracic spine. The mitral valve ring (black arrow) is displaced anteriorly, which produced mitral valve prolapse (white arrow). b) M-mode scan of the left ventricle. Arrows indicate the compression of the left ventricle by the thoracic spine.

Two-dimensional echocardiography revealed a narrowed left ventricular cavity (inflow and outflow) caused by the compression of the lordotic thoracic spine, but not an apparent narrowing of the right ventricular cavity. Furthermore, a mitral valve prolapse was observed with the concomitant anterior displacement of the mitral ring (Fig. 4-a). M-mode echocardiography also showed a narrowed left ventricular cavity (Fig. 4-b). The dimensions of the inflow and outflow tracts changed with respiration concomitant with murmur changes (Fig. 5). Systolic time intervals were obtained as a mean of three successive cardiac cycles in each quiet inspiration and expiration (Table I). In the expiratory phase, Q-IIa (interval from Q-wave of ECG to aortic second sound) and LVET shortened, PEP/LVET increased, and Q-IIp (interval from Q-wave of ECG to pulmonic second sound) was prolonged.

Clinical course: About 1 year and a half later, he died because of the
Fig. 5. M-mode echocardiograms. a) The mitral valve (MV) shows pansystolic bowing (white arrow) and diastolic fine flutterings (black arrow). b) M-mode echocardiogram of outflow tract of the left ventricle. Dimension of the outflow tract changed with respiration.

Table I. Respiratory Changes of Cardiac Cycles

<table>
<thead>
<tr>
<th></th>
<th>Q-IIa (sec)</th>
<th>ET (sec)</th>
<th>PEP (sec)</th>
<th>PEP/ET</th>
<th>Q-IIp (sec)</th>
<th>HR (beat/min)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Inspiratory phase</td>
<td>0.340</td>
<td>0.255</td>
<td>0.085</td>
<td>0.333</td>
<td>0.375</td>
<td>72</td>
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<tr>
<td>Expiratory phase</td>
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<td>0.245</td>
<td>0.085</td>
<td>0.347</td>
<td>0.400</td>
<td>71</td>
</tr>
</tbody>
</table>

Q-IIa=interval from Q-wave of ECG to aortic second sound; ET=ejection time of the left ventricle; PEP=pre-ejection period of the left ventricle; Q-IIp=interval from Q-wave of ECG to pulmonic second sound; HR=heart rate.

progression of respiratory dysfunction and congestive heart failure. He was cyanotic and tachycardiac (heart rate around 130 beat/min) and his arterial blood gas analyses revealed hypercapnia (PCO₂ around 80 mmHg) and hypoxia (PO₂ around 50 mmHg) just before his death. Autopsy was not performed because permission of his family could not be obtained.

DISCUSSION

This patient showed a severe deformation in the thoracic spine and thorax, with a marked depression in the respiratory function (vital capacity
500–600 ml). It has already been reported that the deformation of the thoracic spine and thorax itself causes impairment of the cardiopulmonary function, including hypertrophy of the right ventricle and decreases of the partial pressure of oxygen in the arterial blood.\textsuperscript{10–14} The impairment of the cardiorespiratory function is more prominent in progressive muscular dystrophy.\textsuperscript{1–7}

Cardiac dysfunction due to degeneration of the cardiac muscle has been reported in progressive muscular dystrophy.\textsuperscript{15–22} However, no report has discussed the direct effect which deformities of the thoracic spine and thorax have on the heart. In this patient, lordosis of the thoracic spine was remarkable, with the heart being caught between the thoracic spine and sternum. The inflow and outflow tracts of the left ventricle were compressed and narrowed, and subsequently an early diastolic murmur and systolic murmur occurred. These cardiac murmurs became louder during the expiratory phase when the thorax involuted and the compression on the heart increased. In addition, the systolic murmur was very prominent and accompanied by thrill, and this murmur radiated from the sternum toward the neck. No anomaly of the aortic valve, aortic root or apparent obstruction of the right ventricle was detected by echocardiography. Therefore, although we could not designate invasively the presence of stenosis in the left side of the heart and the absence of stenosis in the right side of the heart, the origin of the murmurs was suspected to be the narrowed inflow and outflow tracts of the left ventricle. It is possible that the murmurs were produced by either of the apparent stenosis or merely by the narrowing (but not stenosis) of the left ventricular cavity. Although Doppler echocardiography is useful to estimate the stenosis noninvasively,\textsuperscript{23–26} Doppler flowmetry was not available in our institution.

In the analysis of cardiac cycles obtained from the carotid pulses, the ejection time and Q-IIa time were shorter during expiration than during inspiration. This is opposite to the responses of normal subjects as reported by Nandi et al.\textsuperscript{27} and Pigott et al.\textsuperscript{28} Moreover, it has been reported that the shortening of the ejection time and Q-IIa time during inspiration compared with expiration is greater in patients with pulmonary disease, constrictive pericarditis or cardiac tamponade than in normal subjects.\textsuperscript{29–31} Therefore, in this patient, it appears that the changes in the cardiac cycles during respiration were not caused by pulmonary dysfunction, but by a narrowing of the left ventricle and left atrium, due to its being compressed by the lordotic thoracic spine. During expiration this compression was augmented, and thus further aggravated the narrowing of the inflow and outflow tracts of the left ventricle.
It is known that the incidence of mitral valve prolapse is high among patients with progressive muscular dystrophy.\textsuperscript{32)-37)} Abnormal contraction of the left ventricle and dysfunction of the papillary muscles due to degeneration of the papillary muscles and myocardium of the left ventricle have generally been suggested as the principal causes.\textsuperscript{33)-35)} However, we have also investigated dysfunction of the left ventricle, lordosis of the thoracic spine and the subsequent flattening of the thorax concerned with the mitral valve prolapse, and have found that the thorax deformity strongly correlated with the prolapse.\textsuperscript{37),38)} Thus, we suggest that this is a typical case of anterior displacement of the mitral valve annulus, markedly from the back, and that the distance between the mitral leaflet and papillary muscles was shortened, which in turn resulted in prolapsing of the mitral valve beyond the mitral annulus, as Cheng suggested.\textsuperscript{39)}

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