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Case Report

Duchenne muscular dystrophy with platypnea-orthodeoxia from Chilaiditi syndrome

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Abstract

Introduction: Chilaiditi syndrome is a rare pathophysiology in which the colon or other organs are interposed between the diaphragm and liver, and respiratory or digestive symptoms sometimes manifest. Although there have been some cases of Chilaiditi syndrome complicating neuromuscular disorders, none have described resulting respiratory or digestive symptoms.

Case presentation: Our patient was a 20-year-old man with DMD who had been receiving noninvasive positive-pressure ventilation during the night. He experienced respiratory distress when changing from a supine to sitting position. Ventilator adjustment did not relieve the respiratory distress. Abdominal computed tomography revealed marked constipation and interposition of the transverse colon between the diaphragm and liver, indicating Chilaiditi syndrome. The right side of the diaphragm was elevated by the interposed transverse colon when the respiratory distress was present on chest radiograph, but not when symptoms were absent. The patient was diagnosed with platypnea-orthodeoxia attributed to Chilaiditi syndrome. The respiratory distress was improved by the relief of constipation, in addition to the usage of the ventilator throughout the day.

Conclusion: The rare symptoms and pathophysiology of DMD complicated by Chilaiditi syndrome are reported and discussed herein.

Keywords: Duchenne muscular dystrophy (DMD); Chilaiditi syndrome; Platypnea-orthodeoxia; Constipation; Respiratory distress

1. Introduction

Chilaiditi syndrome is a rare pathophysiology in which the colon or other organs are interposed between the diaphragm and liver [1]. The syndrome is generally asymptomatic; however, it may cause symptoms such as abdominal pain, vomiting, respiratory distress, and arrhythmia [2–4]. The incidence of Chilaiditi syndrome is 17% in patients with neuromuscular disorders compared with 0.14–0.25% in the general population [3,5]. No reports have described a neuromuscular disorder complicated by Chilaiditi syndrome that manifested with respiratory or gastrointestinal symptoms. The pathogenicity is unknown, although it was not thought

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that Chilaiditi syndrome affected the diaphragm in neuromuscular disorders [5].

2. Case report

The patient was a 20-year-old man diagnosed with Duchenne muscular dystrophy (DMD), deletion of exons 30–43 of the dystrophin gene. He was unable to walk by the age of 9 years, and had used a wheelchair since he was 10 years old. He underwent surgery for scoliosis at 15 years old, and received noninvasive positive-pressure ventilation (NPPV) during the night since he was 18 years old. He had suffered from constipation since he was 18 years old. Respiratory distress was noted when his posture changed from the supine to sitting position 6 months previously. This occurred frequently, particularly when the patient had constipation. We checked his respiratory status and adjusted the NPPV.

On admission, the patient had severe muscle weakness. He could sit and use his motorized wheel chair unaided. His weight was 38 kg, and his general SpO_2 was 98% to 100% when at rest in the supine position. His arterial oxygen saturation (SpO_2) decreased to 78% after posture changed from the supine to sitting position; however, it improved when he returned to the supine position. His systolic blood pressure and pulse were unchanged when the SpO_2 decreased. Arterial blood gases before and after testing changed as follows, indicating respiratory acidosis: pH, 7.405–7.331; partial pressure of carbon dioxide, 38.1–44.3 mmHg; and partial pressure of oxygen, 95.8–86.8 mmHg. The spirometer revealed a decrease in vital capacity from 340 mL in the supine position to 300 mL in the sitting position. The CO_2 monitor during the night showed an average of 71 mmHg, indicating a high pCO_2. At first, we thought that the desaturation and dyspnea when sitting was caused by respiratory muscle fatigue resulting from the respiratory distress at night. The NPPV settings were changed and the patient's night pCO_2 returned to normal; however, his respiratory distress when sitting persisted. Abdominal computed tomography (CT) revealed interposition of the transverse colon between the diaphragm and liver, indicating Chilaiditi syndrome (Fig. 1), along with marked constipation.

We thought that Chilaiditi syndrome might be associated with respiratory distress. Therefore, we used chest radiography at changing his posture from a supine to sitting position, when the desaturation and dyspnea were present or not, respectively. The right side of the diaphragm was elevated by the interposed transverse colon when the respiratory symptoms were present, but not when they were absent (Fig. 2). Thus, the patient was diagnosed with platypnea-orthodeoxia attributed to Chilaiditi syndrome. He began to use the NPPV throughout the day and received laxatives, which improved the respiratory distress. Subsequently, the patient returned to using NPPV only during the night because his constipation and platypnea-orthodeoxia were improved.

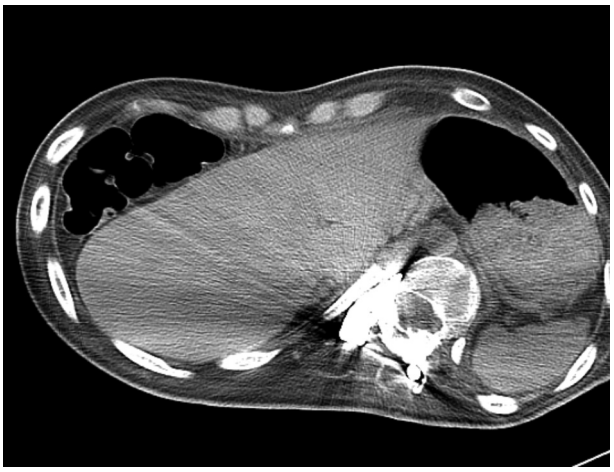

Fig. 1. Abdominal computed tomography (CT); colonic gas was interposed between the diaphragm and liver.

3. Discussion

Reports have described respiratory distress in patients with neuromuscular disorders by analyzing the difference in vital capacity between various positions. In DMD, there was no significant difference between the vital capacity while sitting and supine [6]. However, a decrease in SpO_2 and respiratory distress frequently occurred when our patient changed from supine to sitting. Platypnea-orthodeoxia is a rare condition in which hypoxemia and respiratory distress are caused by changing the posture from the supine to the standing or sitting position; it is relieved by returning to the supine position [7,8]. It can be caused by a right-to-left shunt in the heart. However, platypnea-orthodeoxia may result from a ventilation-perfusion imbalance when sitting because of anatomical abnormalities, e.g., spinal or thoracic deformities, pulmonary emphysema, or pneumonia [8]. In our case, no underlying diseases that could cause platypnea-orthodeoxia were detected. However, abdominal CT revealed Chilaiditi syndrome that might be associated with platypnea-orthodeoxia.

Chest radiography confirmed elevation of the right diaphragm in the sitting position at the onset of platypnea-orthodeoxia, whereas the diaphragm was not elevated in the absence of platypnea-orthodeoxia. The internal organs are pulled down by gravity, which expands the thoracic cavity by contracting the diaphragm and increasing functional residual capacity when sitting [9]. The opposite phenomenon was observed in our case. The following events likely