Yonsei Medical Journal
Vol. 39, No. 2, pp. 175~179, 1998

☐ Case Report ☐

Acute Gastroparesis in Duchenne's Muscular Dystrophy

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Duchenne's muscular dystrophy(DMD) is an X-linked recessive disease. Clinical descriptions of the disorder focus principally on skeletal muscle degeneration. Another manifestation, which involves the gastrointestinal tract, may be fatal. But its prevalence remains undefined. We report here a case of acute gastroparesis associated with Duchenne's muscular dystrophy. In our case, the patient's symptoms were improved by prokinetic agents and timely decompression in life-threatening acute gastric dilatation.

Key Words: Duchenne's muscular dystrophy, gastroparesis, prokinetic agents

Duchenne's muscular dystrophy(DMD) is a fatal X-linked recessive disease and the most common congenital neuromuscular disorder of childhood (Emery, 1991). Clinical descriptions of the disorder focus principally on skeletal muscle degeneration. Muscular weakness is inevitably progressive, leading to the complete inability to ambulate by 9 to 11 years of age (Allsop and Ziter, 1981). Another manifestation, frequently overlooked, involves the gastrointestinal tract. Although gastrointestinal involvement in DMD has previously been documented, its prevalence remains undefined (Robin *et al.* 1963; Leon *et al.* 1986; Jaffe *et al.* 1990). The symptoms, which include bloating, a feeling of fullness and in some cases a catastrophic syndrome of acute gastric dilatation and intestinal pseudo-obstruction, may be fatal (Barohn *et al.* 1988). It may be produced by abnormalities of the smooth muscle or myenteric plexus (Verne and Sninsky, 1995). We describe here a case of acute gastroparesis associated with Duchenne's muscular dystrophy.

Received November 18, 1997
Accepted January 19, 1998
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CASE REPORT

A 14-year-old boy with Duchenne's muscular dystrophy was admitted to the hospital for vomiting and abdominal pain of 2 days' duration. He had no previous history of pulmonary tuberculosis, hepatitis, diabetes mellitus, or hypertension. He had been diagnosed with muscular dystrophy at 2 years of age due to a gait disturbance. Throughout his life, the patient had suffered from recurrent attacks of abdominal pain associated with generalized abdominal distension, nausea and vomiting, but not from any episode of pneumonia or acute respiratoy failure.

On admission, he was suffering from anorexia, vomiting and abdominal pain. The pain was described as sharp and constant. He had been unable to eat or drink for 2 days. He denied having fever,

chills, dyspnea, cough, heartburn or melena, but he did complain of constipation.

On physical examination, his height was 150cm, weight was 18kg, blood pressure was 100/70mmHg, and body temperature was 36.7℃. The patient appeared ill. Skin turgor was normal. His sclera were anicteric. Chest excursion was asymmetric with shallow inspiration. Lungs were clear to auscultation. No cardiac rub or murmur was detected. The abdomen was mildly distended but soft, bowel sounds were hyporeactive and no high-pitched tinkles or rushes were heard. There was direct tenderness in his abdomen with no rebound tenderness or guarding. All extremities were dystrophied and contracted. Laboratory investigation demonstrated a white blood count of 12,910/uL with 92.1% neutrophils, 6.7% lymphocytes, and 1.2% monocytes. Hemoglobin (14.9 g/dL), electrolytes (Na 141 mM/L, K 4.2 mM/L, Cl 102 mM/L, bicarbonate 25 mM/L), BUN (14.9 mg/dL), creatinine (0.3 mg/dL), amylase (8 1IU/L) lipase (70 IU/L), T. protein (6.9 g/dL), albumin (4.7 g/dL), calcium (9.2 mg/dL), inorganic P. (4.3 mg/dL), uric acid (2.7 mg/dL), cholesterol (120 mg/dL), AST (78 IU/L), ALT (69 IU/L), T. bilirubin (1.8 mg/dL), and alkaline phosphatase (124 IU/L) were obtained. Urinalysis was positive for bilirubin with a specific gravity of 1.015. Microscopic analysis was normal. Radiograph of the chest showed a calcified nodule in the left hilar area. An electrocardiogram showed a sinus tachycardia at a rate of 112, with an incomplete RBBB. There was no free air or evidence of mechanical obstruction on

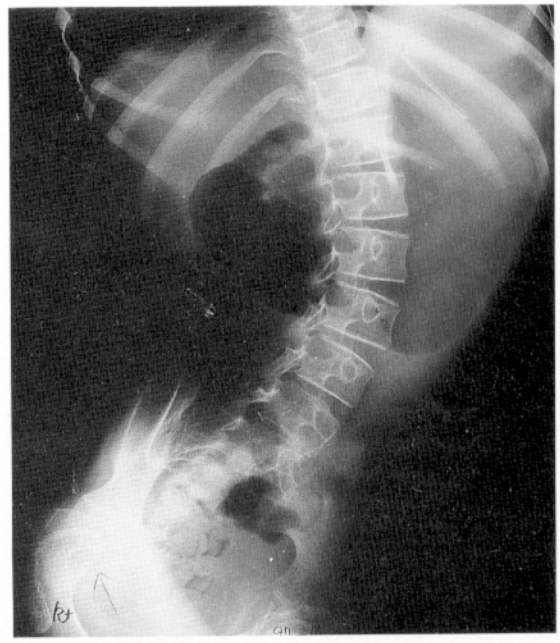

Fig. 1. Simple abdomen shows a markedly dilated stomach.

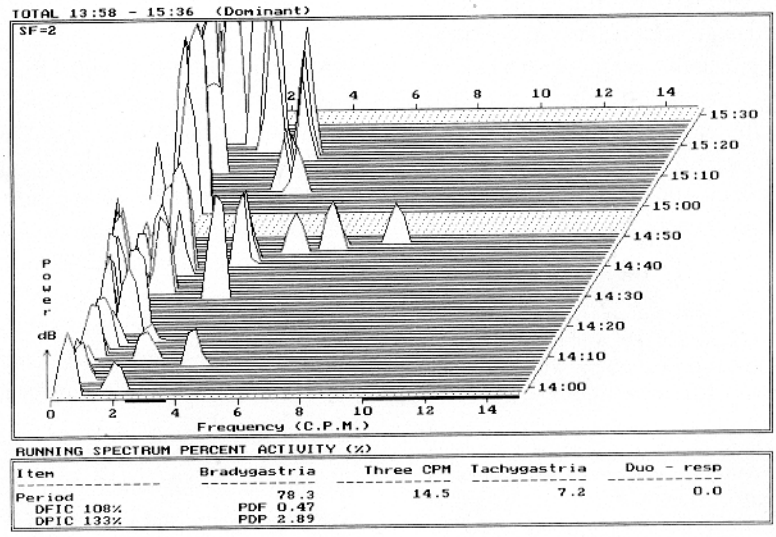

Fig. 2. Electrogastrogram shows a marked bradygastria.

plain abdominal radiograph. But it showed a markedly dilated stomach (Fig. 1).

On suspicion of having a motility disorder of the gastrointestinal tract, gastric myoelectrical activity was recorded by cutaneous electrogastrography (EGG; Digitrapper EGG, Synectics, Stockholm, Sweden) and it revealed a marked bradygastria (Fig. 2). Gastric emptying was assessed by a gelatin capsule containig 24 ring-shaped (1×4.5 mm) radio-opaque markers (ROM; Sitzmarkers®, Fortworth, Texas, USA). After overnight fast, a standard test meal was administered with ROMs (Park *et al.* 1997). A supine plain abdominal radiograph was taken 12, 24 and 48 hours after ingestion of the test meal. The mean emptying of ROMs was 60% at 24h and it showed a delayed clearance (Fig. 3). Under the diagnosis of gastric hypomotility, intravenous erythromycin 100 mg and metochlopramide 7.5 mg were administered daily with gastric decompression. Thereafter his symptoms were improved and he was discharged after two weeks.

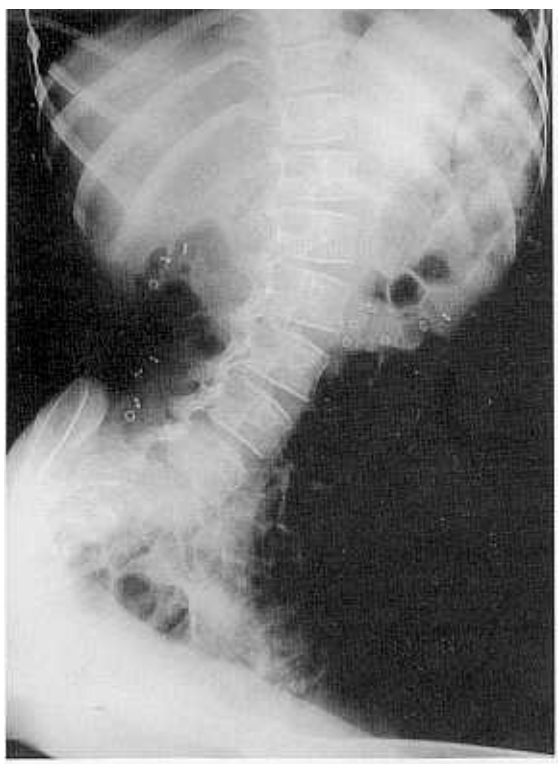

Fig. 3. A supine plain abdominal radiograph was taken 24 hours after ingestion of the ROMs. The mean emptying of ROMs was 60% at 24h and showed a delayed clearance.

DISCUSSION

Duchenne's muscular dystrophy is an X-linked disorder that causes skeletal and cardiac muscle degeneration leading to progressive weakness and death before the end of the third decade of life. It has an incidence of approximately 30 per 100,000 newborn males. By age 16 to 18, patients are predisposed to serious, sometimes fatal pulmonary infection. Other causes of death include aspiration of food and acute gastric dilatation. A cardiac cause of death is uncommon despite the existence of a cardiomyopathy in almost all patients (Willig, 1994).

Although gut involvement in DMD is currently thought to be due to smooth muscle degeneration, neurogenic defects cannot be excluded. The gene that results in DMD was recently isolated and it was localized to the short arm of the X chromosome at the Xp21 site. DMD is caused by a mutation of the gene responsible for producing dystrophin (Burghes *et al.* 1987; Koenig *et al.* 1987). At the present time, the physiologic function of dystrophin remains unclear. It is localized on the surface membrane of normal muscle and cardiac muscle fibers, as well as smooth muscle fibers (Bonilla *et al.* 1988). Dystrophin plays an important physiological or structural role in the conduction system and seems to affect cell motility, regulation of cell shape, and intracellular transport (Miyatake *et al.* 1991).

We describe here a case of acute gastroparesis associated with DMD, but the overall prevalence is unknown. Crowe reported clinical gastric dilatation for the first time in the English literature in a symptomatic 9-year-old boy with DMD (Crowe, 1961). Several other studies describe various GI manifestations of DMD (Staiano *et al.* 1992; Bensen *et al.* 1996). The symptoms tend to appear, subside, and reappear in recurring episodes. The usual presentation is generally related to abdominal distension.

Barohn *et al.* showed objective evidence of functional smooth muscle impairment in Duchenne's dystrophy with radionuclide gastric-emptying studies

(Barohn *et al.* 1988). The radio-opaque marker technique is also a non-invasive method for the study of gastric emptying. The ROMs assess gastric emptying of indigestible solids and reflect the onset of the interdigestive migrating motor complex (Feldman *et al.* 1984; Chang *et al.* 1996). The mean emptying of ROMs averaged 60.2% at postprandial 3h (Park *et al.* 1997). In this patient, the mean emptying of ROMs was 60% at 24h after ingestion and it suggested a delayed clearance of indigestible solid meals. Electrogastrography is usually referred to as the non-invasive technique of recording gastric myoelectrical activity by placing electrodes on the abdomen. The dominant frequency of the EGG reflects the frequency of the gastric slow wave. The gastric dysrhythmias are believed to be associated with gastric motility disorders (Telander *et al.* 1978; Chen and McCallum, 1993). The electrical abnormalities were obtained by Borrelli *et al.* in children with progressive muscular dystrophy (Borrelli *et al.* 1997). The patient in the present study showed a marked bradygastria.

Acute gastroparesis can be caused by a variety of disease, and for simplicity it is divided into myopathic and neuropathic categories. Recently, it has been suggested that neurogenic defects can underlie gastric dysmotility in DMD (Miyatake *et al.* 1991; Borrelli *et al.* 1997). The patients who have myopathic disease usually do not respond to prokinetic agents such as cisapride and erythromycin. However, those with neuropathic disease will usually show a response (Colemont and Camilleri, 1989). The use of prokinetic agents to decrease gastrointestinal symptoms in DMD is worthy of investigation. In this study, the patient showed a response to erythromycin that stimulated antral activity.

In our case, we believe that the patient's symptoms were improved by prokinetic agents. With the increased awareness of upper gastrointestinal motility disorders in DMD, timely decompression in life-threatening acute gastric dilatation and prokinetic agents should be tried in an attempt to restore normal gastrointestinal propulsion.

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