

[ CASE REPORT ]

## Achalasia in a Patient with Myotonic Dystrophy

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### Abstract:

Myotonic dystrophy is a progressive disorder mainly affecting the voluntary muscles. We herein report a rare case of myotonic dystrophy complicated with high-resolution manometry-defined achalasia, the pathology of which is absent relaxation of the smooth muscles of lower esophageal sphincter (LES). In the present case, achalasia was considered a complication of myotonic dystrophy instead of sporadic achalasia, as on performing high-resolution manometry, the finding of an impaired LES relaxation (myotonic phase) changed to a totally emaciated LES function (muscle weakness phase) as myotonic dystrophy progressed.

**Key words:** myotonic dystrophy, achalasia, high-resolution manometry, lower esophageal sphincter, smooth muscle, voluntary muscle

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### Introduction

Myotonic dystrophy is a progressive disorder that mainly affects the voluntary muscles; the systemic skeletal muscles are unable to relax (myotonic phase) and gradually become emaciated (muscle weakness phase) (1, 2). Other complications, such as cataracts and cardiomyopathy, are also seen (3).

### Case Report

A 54-year-old woman who had been diagnosed with myotonic dystrophy 18 years earlier was referred to our unit because of chronic cough and anorexia. On performing gastroscopy, no obstructive findings were observed. High-resolution manometry (HRM) revealed absent relaxation of the lower esophageal sphincter (LES; yellow triangle, Fig. 1A) and no normal peristalsis (white arrow), resulting in a diagnosis of type I achalasia being made. No pressurization was observed in the upper esophageal sphincter (UES; red triangle). Timed barium swallow showed some stagnant barium remaining in the esophagus (Fig. 2), and computed tomography showed a dilated esophagus (Fig. 3).

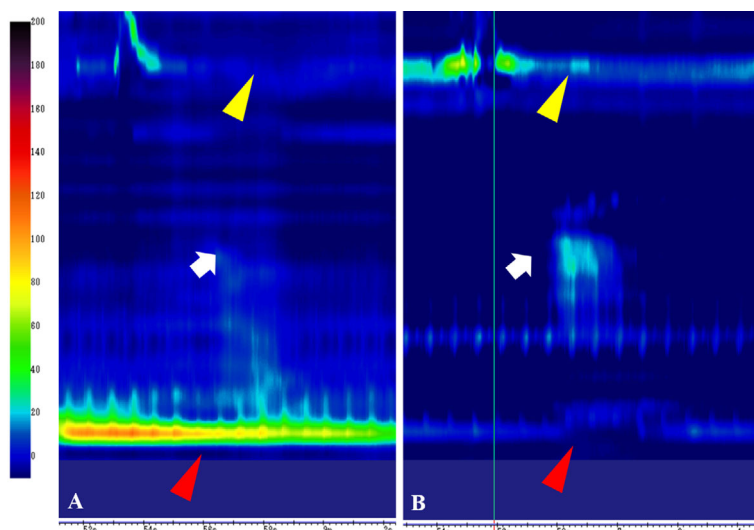
The patient was not provided any treatment because her esophageal symptoms were still mild at the time, and her severe systemic symptoms were the primary concern, rather

than her esophageal symptoms. After three years, she experienced dysphagia and regurgitation, so HRM was performed again, revealing a finding of aperistalsis with failure of the LES function (Fig. 1B).

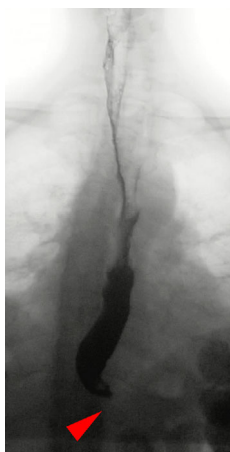
### Discussion

Dysphagia in cases of myotonic dystrophy has been reported for quite some time. Its causative mechanism was considered to be myotonia or weakness of the striated muscles from the pharynx to the upper third of the esophagus; therefore, it is also termed cricopharyngeal achalasia (4-7). To our knowledge, this is the first report of myotonic dystrophy complicated with HRM-defined achalasia, the pathology of which is absent relaxation of the smooth muscles of the LES. Furthermore, achalasia in the present case was considered to be a complication of myotonic dystrophy instead of sporadic achalasia, as on performing HRM, the UES function declined and the finding of no relaxation of LES (myotonic phase) changed to a totally emaciated LES function (muscle weakness phase) as myotonic dystrophy progressed. Therefore, achalasia as a complication of myotonic dystrophy should be followed up, but not necessarily for achalasia.

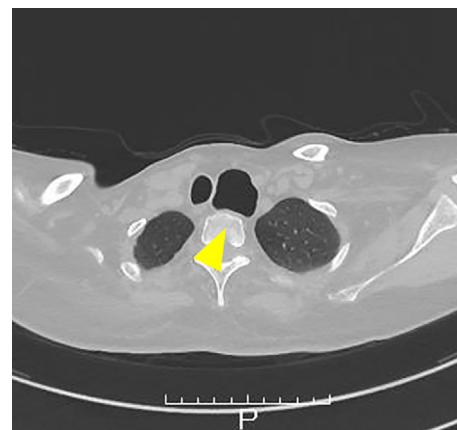
**The authors state that they have no Conflict of Interest (COI).**



**Figure 1.** A: High-resolution manometry (HRM) revealed absent relaxation of the lower esophageal sphincter (red triangle; the mean LES pressure was high, 30.1 mmHg), no upper esophageal sphincter pressure (yellow triangle), and no normal peristalsis (white arrow), resulting in a diagnosis of achalasia. B: HRM finding three years later, showing aperistalsis without LES pressurization.



**Figure 2.** Esophagography revealed a “bird-beak” appearance of the barium, indicating achalasia.



**Figure 3.** A computed tomography scan showing the dilated esophagus.

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