
Mihai-Mirel STOICESCU; Mihai MOCANU; Felicia GALOS; Mihai MUNTEANU; Simina VISAN; Coriolan ULMEANU; Mihaela BALGRADEAN

a"Carol Davila” University of Medicine and Pharmacy, Bucharest, Romania
b”Marie Sklodowska Curie” Emergency Children’s Hospital, Bucharest, Romania

ABSTRACT

We report the case of a rare association between achalasia and Down syndrome in a child presenting with symptoms that suggest a gastroesophageal reflux. Evaluation of the patient with 24-hour multichannel intraluminal impedance and pH recording and upper endoscopy lead to the diagnosis of achalasia. However, the persistence of the symptoms after the concurrent surgical myomectomy and fundoplication has led to repeat pH-impedance monitoring testing and endoscopy, which identified the presence of gastroesophageal reflux disease. We emphasize in this paper the importance of multichannel intraluminal impedance and pH monitoring in detecting esophageal motility disorders.

INTRODUCTION

Combined multichannel intraluminal impedance and pH recording (MII-pH) in the esophagus has been available for more than fifteen years for the study of fluid movement in the esophagus. It reliably detects solid, liquid, gas and mixed liquid-gas reflux episodes in suspected cases, and correlates them with concurrent pH measurements. MII-pH allows to detect all episodes of reflux regardless of its content (liquid, gas, and mixed) and its pH, that is to recognize episodes of acidic reflux, weakly acidic reflux, and alkaline reflux. Because impedance catheters have multiple sets of impedance-measuring rings, bolus movement and direction (antegrade or retrograde) can be assessed (1).

Normal standards for MII-pH are not yet established in children, but it allows one to increase the accuracy of observations during esophageal pH-monitoring and draw relevant conclusions (2).

In this paper we present a case of rare association of achalasia and gastroesophageal reflux disease (GERD) in a child with Down syndrome and comorbid conditions, where the combined multichannel intraluminal imped-
ance-pH recording was useful in establishing the diagnosis and monitoring the outcome of surgical and medical therapy.

CASE REPORT

A 6-year-old male child was referred to our Department of Pediatric Surgery accusing the following:
- Chronic vomiting
- Occasional head dystonia (opistotonus)
- Food refusal
- Failure to thrive.

Symptom onset was loosely estimated by the mother to the age of four.

The child was known to have Down syndrome and Legg–Calvé–Perthes disease, the latter being managed with applied orthoses that restricted the sitting position.

The patient was referred to our Department of Gastroenterology and, based on clinical presentation and history, the child was initially diagnosed with gastroesophageal reflux and proton pump inhibitors were prescribed.

One month later, the lack of response to drug therapy prompted us to perform esophageal MII-pH monitoring and an upper endoscopy to assess the degree of reflux and the presence and nature of esophagitis (GERD esophagitis, eosinophilic esophagitis).

A Digitrapper® pH-Z equipment (Given Imaging, Israel) was used to monitor pH and impedance according to the standard protocol. A pediatric-sized combined 6-channel impedance pH-metry catheter was placed transnasally.

The initial position of the pH sensor was calculated according to the Strobel formula and a chest X-ray performed to check the accuracy of the placement.

The child’s mother was asked to keep a diary during the whole monitoring, recording the timing of meals and the occurrence of symptoms, which was later transferred to the digital recording.

Due to the inadequate accuracy of current software analysis, the tracing was manually reviewed by a human operator.

Contrary to the expectations, the MII-pH tracings demonstrated low baseline impedance levels in the distal channels, which can occur both in chronic esophagitis and achalasia (Figure 1).

The next day, the patient was sedated and an upper endoscopy was performed showing a lax, dilated esophagus with fluid residues, absence of propulsive contractions, resistance to cardial intubation and mucosal changes due to food retention (yellowish deposits, thickening of the mucosa) (Figure 2).

A diagnosis of achalasia was made, further confirmed by a barium swallow the following day.

Drug therapy was stopped and in the next weeks the child underwent a laparoscopic
Heller myotomy with Toupet fundoplication, with full postoperative recovery. Postoperatively, the child was given proton pump inhibitors for three months.

At the 2 months follow up the patient was symptom free and weight gain was restored, but at three months post procedure, vomiting and lack of appetite reappeared, later followed by neck dystonia, all of which raised concerns regarding a possible myotomy failure.

Therefore, a new esophageal MII-pH was scheduled and performed after a seven-days drug-free period. The new tracing demonstrated this time low to normal baseline impedance values and an increased number of reflux episodes, indicating the presence of a gastroesophageal reflux disease.

A second upper endoscopy revealed mucosal erythema of the distal esophagus and small, less than 5 mm long, nonconfluent erosions (Grade A esophagitis according to the Los Angeles classification). Multiple mucosal biopsies from the distal esophagus confirmed the diagnosis of GERD esophagitis.

The child was given proton pump inhibitors and a sodium alginate preparation for three months. Two months later, the symptoms had abated and a repeat upper endoscopy revealed complete microscopic mucosal healing of the esophagitis. 

**DISCUSSION**

Achalasia is a rare esophageal motor disorder, linked in some patients to markers on chromosome 12 q13 and inherited in an autosomal recessive manner. It has been associated with other conditions such as Allgrove Syndrome (achalasia, adrenocorticotropic hormone insensitivity, alacrima) (3), familial visceral neuropathy, achalasia microcephaly syndrome, Parkinson’s disease and depressive disorders (4,5). The illness is a rare condition in children, with an estimated incidence in general population of 1 case per 10,000, of which fewer than 5% manifest symptoms before the age of 15 years. The mean age at the time of diagnosis in the pediatric patients, taken from a recompilation of all of the pediatric series available was 8.8 years (6).

Achalasia presents as a functional obstruction at the esophagogastric junction characterized by partial or incomplete lower esophageal sphincter relaxation and a lack of esophageal peristalsis.

On the other hand, gastroesophageal reflux is the most common condition associated with chronic vomiting and failure to thrive in the pediatric population, with a peak incidence in infancy and adolescence. It tends to subside after 12 months of age, becoming increasingly rare in preschool and school aged children.

Therefore, the initial presentation in our case report suggested a gastroesophageal reflux, and the initial treatment was aimed at this diagnosis. It was as the treatment failed to show results that further investigations were performed.

Several other reasons led us to believe the symptoms are due to gastroesophageal reflux only: the presence of Down’s syndrome, the mental impairment of the patient and the neck dystonia (suggesting a Sandifer syndrome).

After performing the surgical cure of achalasia and the symptoms’ persistence, several possible explanations arose to account for this: persistence of achalasia (surgical failure), gastroesophageal reflux as a complication of surgery or the presence of a gastroesophageal reflux unmasked by the removal of the motor esophageal obstacle.

This is where the pH-impedance monitoring became a useful diagnostic tool, ruling out the persistence of achalasia in our patient and detecting an abnormal number of gastroesophageal reflux episodes.

Multichannel intraluminal impedance combined with pH monitoring records esophageal events with a probe placed transnasally and connected to a portable data recorder. The method allows the detection of reflux based on changes in resistance to the electrical current flow between several electrodes alongside the catheter, when the bolus moves between them. The direction of the bolus movement is made possible by the simultaneous recording at different levels. Combined pH-MII catheters are available for every age group of pediatric patients, from infants to adolescents (7).

The presence of a low baseline impedance in the tracings has been commonly associated with mucosal inflammation related to esophageal acid exposure and hypothesized to be a marker of microscopic changes of the esophageal mucosa (8,9). However, the same low baseline impedance levels are associated with achalasia, low impedance values reflecting chronic fluid retention in the lower esophagus (10,11).
Early in the evolution of achalasia, the esophagus may show minimal change or appear to be normal, and patient’s presentation may mimic GERD. Characteristic endoscopic and barium swallow changes occur later, thus delaying the diagnosis. Combined impedance and pH monitoring of the esophagus might detect achalasia long before these changes become obvious, based on detection of a low baseline impedance in the distal channels, absent bolus clearance and air presence in the upper esophagus (detected as gas by the impedance sensors) (5).

In our patient’s case, the combined pH and impedance monitoring was especially valuable because it offered the first clue to the diagnosis of achalasia and later detected the presence of gastroesophageal reflux secondary to surgery. Careful interpretation of low baseline tracings obtained from patients that undergo esophageal pH-impedance monitoring might be able to detect early cases of achalasia.

Conflicts of interests: none declared.
Financial support: none declared.

REFERENCES