



Eating disorder or oesophageal achalasia during adolescence: diagnostic difficulties

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Abstract

Marine was a fourteen and a half-year-old adolescent female hospitalized for an eating disorder (ED) of the anorexic type with purging behaviors. She has had a complicated life course, made up of disruptions and discontinuities, both family and school. Since the age of five, Marine had been intermittently treated in psychiatry for a diagnosis of oppositional defiant disorder. The current illness started with spontaneous and induced vomiting associated with major weight loss (body mass index, 15.27 kg m⁻²). The diagnosis of anorexia nervosa was established after several opinions from professionals in five Parisian university pediatric departments, where additional investigations were carried out without any somatic cause being identified. In this context, Marine was transferred to a child psychiatry unit. There, she had acute dyspnea during the insertion of a nasogastric tube. As a result, a new specialized opinion was sought from a pediatric gastroenterologist and further explorations were performed (oeso-gastrointestinal transit and manometry), leading to the conclusion to an oesophageal achalasia requiring surgical treatment. This case report highlights that the exclusion of any organic disorder should be a priority in the diagnostic assessment of an ED. Oesophageal achalasia is a rare differential diagnosis and should be considered in case of swallowing difficulties or dysphagia. Health care professionals should take care to provide appropriate somatic follow-up for patients with psychiatric disorders.

Keywords Achalasia · Adolescence · Anorexia nervosa · Eating disorder

Introduction

Marine was a 14½-year-old adolescent female consecutively treated in five different pediatric units over 9 months for dysphagia with vomiting associated with weight loss, leading to the diagnosis of anorexia nervosa.

Case report

Up to the age of three, Marine was brought up by her single mother in a community center for women in psychosocial difficulty. From the age of eight, she alternated brief placements in shelters for children at risk or in foster families with living with her mother. Marine's school history had been chaotic from early childhood. Since the age of five, she had received intermittent psychiatric treatment with a diagnosis of oppositional defiant disorder and then substance use disorder (tobacco and cannabis). All psycho-educational measures were undermined by Marine and her mother who had herself been diagnosed with a borderline personality

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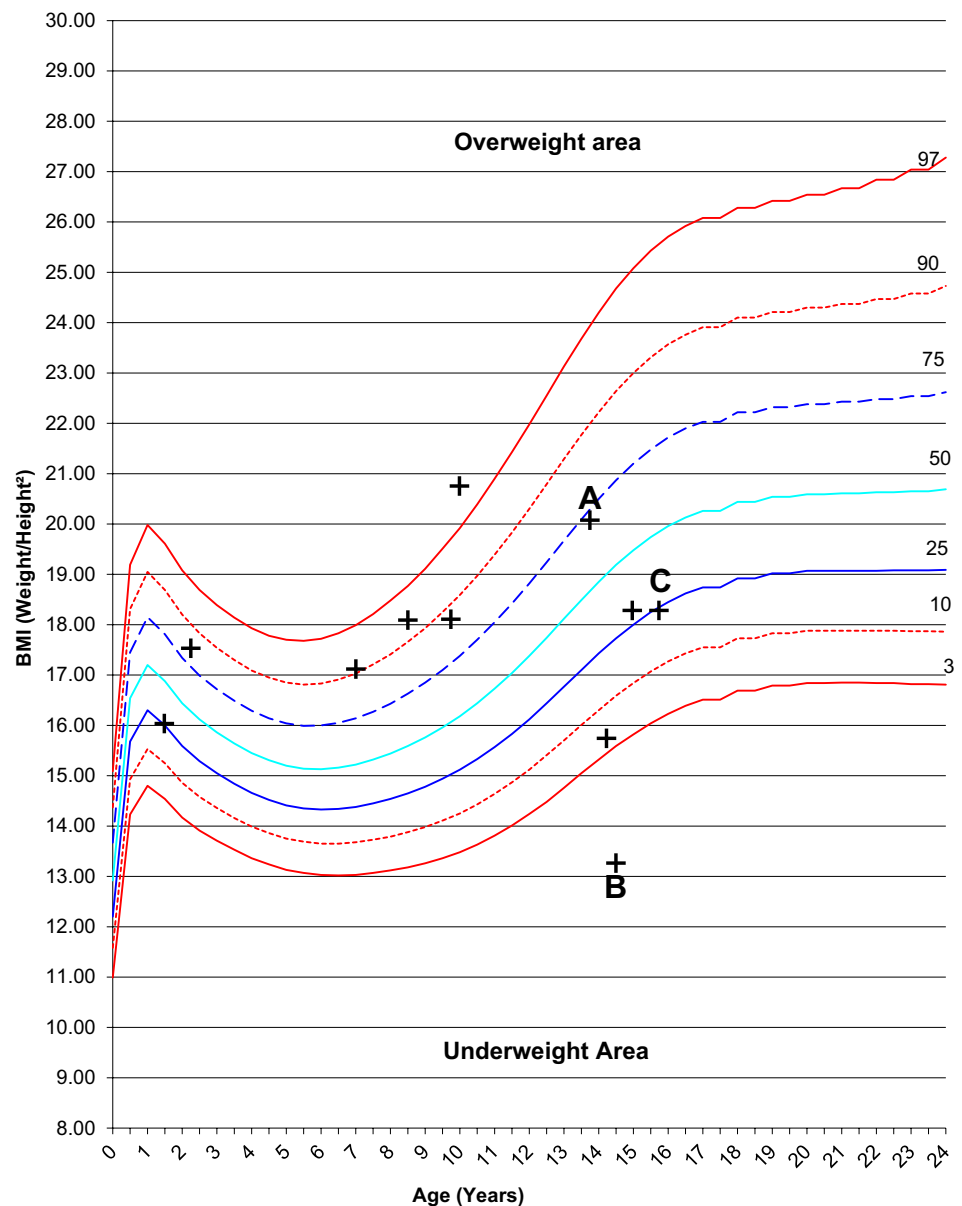
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disorder. Marine's current illness started in the spring (at the end of March) with dysphagia and vomiting, in the absence of any precipitating event, and with no improvement following symptomatic treatment prescribed by the general practitioner. This led to dissatisfaction on the part of Marine's mother who initiated iterative visits to three different emergency units. Somatic investigation included cerebral scan and abdominal echography during the first admission to an emergency unit in the summer (June), then oeso-gastroduodenal fibroscopy in July, all with normal results. Following this, Marine lost 14 kilograms within a month and a half, leading to a body mass index (BMI) of 15.27 kg m^{-2} (see Fig. 1) and secondary amenorrhea in the fall (end of September). The vomiting episodes persisted, occurring at meal times or immediately afterwards, either

spontaneous or self-induced. Marine was admitted to a pediatric gastroenterology unit and then to a pediatric unit as a result of weight loss and hypokalemia (October). In the unit, Marine's mother exhibited violent and maladaptive behavior leading to legal measures. When Marine returned home, the symptoms persisted, including vomiting, and she was again admitted to the pediatric units of two hospitals for several weeks (November). On the basis of the rapid loss of weight, the presence of self-induced vomiting, the active refusal of weight gain, the absence of somatic findings and amenorrhea, the diagnosis of anorexia nervosa, binge-eating/purging type (Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition) was suggested, and a transfer to a psychiatric facility was indicated. The possible disturbance in the patient's body image was difficult to assess at this time

Fig. 1 Patient's BMI growth curve relative to the normative BMI curves based on data from the French CIE-INSERM growth study. Gradations are in centiles. *A* BMI at onset of digestive symptoms, *B* BMI at admission to psychiatry unit, *C* BMI at discharge from psychiatry unit, *CIE* Centre International de l'Enfance, *INSERM* Institut National de la Santé et de la Recherche Médicale



because of oppositional attitudes and reluctance to express her feelings.

At admission to psychiatric unit in December, Marine appeared as severely malnourished (BMI, 13.6 kg m⁻²), with bradycardia. Her condition required refeeding with a nasogastric tube because she was refusing to eat. The chest X-ray was normal. Marine exhibited symptoms of anxiety, intolerance to frustration, and violent clastic behaviors. An anxiolytic pharmacological treatment was initiated, with only partial improvement. During individual interviews, Marine admitted to a long-standing history of binge–purging episodes occurring at home when she was alone since the age of around 10. She could compulsively eat large quantities of sweets, especially in front of the TV, accompanied by purging behaviors. Similar episodes of binge-eating and vomiting recurred during hospitalization. Vomiting of nasogastric refeeding products was regularly observed, but denied by the patient. Further to this, psychometric assessments were carried out, and showed fairly low global intellectual skills, with a IQ score of 74 estimated on the Wechsler Intelligence Scale for Children, WISC-IV (heterogeneous average of the different components). One day, Marine experienced an episode of acute dyspnoea when her nasogastric feeding tube was set up. The emergency chest X-ray was normal. However, a medical consultation was requested following the event, and on account of the fact that the patient, who had resumed normal feeding, was observed jumping to her feet after meals «to force the food down because it is blocked», she said. The consultant pediatric gastroenterologist immediately considered the diagnosis of oesophageal achalasia, which was confirmed by oeso-gastrointestinal transit examination and manometric assessment, and further evidenced by oeso-gastrointestinal projection endoscopy. The patient subsequently underwent surgery with Heller myotomy and anterior anti-reflux mounting.

To date, Marine's somatic progress has been favorable. She has a better nutritional condition despite a few persistent vomiting episodes. Her weight is stable and in the healthy range. However, the psychiatric follow-up is irregular and scant, since the therapeutic alliance has been hindered by diagnostic uncertainties and delays. Recently, for this case report (i.e., 3 years after surgery), the patient's mother was contacted. She confirmed the global improvement in the clinical state of her child but she reported the persistence of vomiting episodes.

Discussion

Oesophageal achalasia is a rare condition in children and adolescents, with an incidence of 0.11/100 000 [1, 2]. Its etiology is most often idiopathic [3], but rare hereditary cases exist, with autosomic recessive transmission (triple

A syndrome: achalasia, alacrymia, and adrenal insufficiency) [4].

Achalasia is a primary motor disorder of the esophagus characterized by the absence of oesophageal peristalsis with hypertonia of the inferior oesophageal sphincter (IOS) and the absence of IOS relaxation following deglutition. This pathology manifests itself mainly with dysphagia for solid and liquid food, regurgitation of undigested aliments, and retrosternal pain. The diagnosis is based on personal history detailing the quality of deglutition and the characteristics of vomiting episodes, an oeso-gastrointestinal transit investigation to detect megaesophagus or food stasis, and the assessment of oesophageal motility using manometry. Current therapeutic options are pneumatic dilation, surgical myotomy, and pharmacological agents intended to decrease pressure at the IOS and facilitate oesophageal emptying [1, 2, 5].

The differential diagnosis between achalasia and an eating disorder (ED) is difficult and a rare occurrence: 36 cases of misdiagnoses have been reported in 26 published studies in the literature [6]. Indeed, the diagnosis of an ED of the anorexic type is often reached in case of weight loss with vomiting. In addition, motor disorders of the esophagus or upper gastrointestinal symptoms (defective gastric filling, bloating and postprandial feeling of satiety) can be seen in patients with anorexia nervosa and bulimia nervosa [6]. Achalasia is a rare disorder, typically starting with dysphagia. The diagnosis is often reached several years after onset, when the symptoms worsen, with spontaneous or induced vomiting and the resulting ionic imbalance, food restrictions and weight loss that can be considerable [6, 7]. This sequence can lead to achalasia being misdiagnosed as an ED. Some studies suggest there is an atypical ED presentation in patients with a retrospective diagnosis of achalasia, with a relationship to food that is less disturbed than in anorexia nervosa, and the absence of an imperative drive to lose weight [6, 7]. Finally, a preexisting ED could be complicated by secondary oesophageal achalasia [8].

Therefore, excluding any organic disorder should be a priority when diagnosing an ED. Oesophageal achalasia should be ruled out in case of difficulty swallowing or dysphagia. In the current case report, the diagnosis of achalasia may have been masked by the complex situation, with an unstable family environment interfering with the patient–doctor relationship, and the patient's personal history of psychiatric disorders.

The difficulties in diagnosing somatic complications occurring in patients with ED have been described [9]. Health professionals should be attentive to these issues and provide adequate somatic follow-up for patients diagnosed with a psychiatric disorder. It is important to ensure a multidisciplinary approach (including internists and psychiatrists)

to the diagnosis and treatment of complex cases with interwoven somatic and psychiatric features [10].

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Compliance with ethical standards

Conflict of interest On behalf of all authors, the corresponding author states that there is no conflict of interest.

Ethical approval All procedures were performed in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Informed consent Consent from the mother of the patient (minor) described in the clinical case was obtained for the sharing of the patient's data in accordance with anonymity requirements.

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