Case report

A case of massive gastric necrosis in a young girl with Rett Syndrome

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Abstract

This is the unusual case of a 17-year-old girl affected by Rett Syndrome (RS) who suffered acute abdominal distension and constipation for a week. Laparotomy showed massive gastric dilatation, with total necrosis and perforation. Total gastrectomy and Y-Roux esophagojejunostomy were performed. We believe the clinical status was caused by the mechanism of air swallowing, present in our patient and typical in RS. In fact, as reported, massive air bloat may result in a decrease of the intramural blood flow with consequential ischemia of the gastric wall. We stress the importance of early detection of the gastroenterological symptoms in these patients, with timely positioning of nasogastric tube and gastrostomy, to prevent serious complications potentially life-threatening as massive gastric necrosis.

Keywords: Rett Syndrome; Abdominal distention; Gastric necrosis; Gastrectomy; Air swallowing

1. Background

Why an organ with a very rich blood supply as the stomach can develop a massive necrosis? Some reports of gastric necrosis from various etiologies as vascular causes, bulimia, selective vagotomy, abdominal trauma, infectious disease, gastric volvulus, intrathoracic herniation, necrotizing gastritis, ingestion of caustics and acute gastric dilatation have been described [1,2]. However, about half the cases seem to be related to large meals and acute gastric dilatation. Thus in literature we found numerous correlations between acute gastric dilatation and neuropsychiatric syndromes as psychogenic polyphagia, anorexia nervosa and Prader-Willi syndrome [2] but only one case of gastric perforation has been described in 1997 in a patient with Rett Syndrome (RS) [3]. This syndrome, first recognized by Andreas Rett in 1966, is a rare, profoundly disabling neurodevelopmental disorder affecting predominantly females, with a prevalence of 1 in 10,000 girls. The patients usually exhibit autistic behaviours, characteristic stereotyped movement, mental retardation, breathing dysfunction and seizures [4].

The aim of this report is to describe a case of a massive gastric necrosis in a girl affected by RS, underlining the importance of the timely monitoring of arising gastro-intestinal symptoms to prevent severe life-threatening complications.

2. Case report

A 17-year-old female with RS was admitted with a 7-day-history of constipation, abdominal distension, vomit and intermittent episodes of hyperventilation with breath-holding. Since the age of 4, the patient presented the typical hand-washing movements and autistic behaviours so the diagnosis of RS was performed in a specialized Neurological Hospital. She was treated with Carbamazepin 900 mg/day for seizure.

Upon arrival, the girl was somnolent, pale, tachypnoic, with marked hypotension (60/40 mmHg), tachycardia (160 beats/min) and seizures. Upon physical examination she had a paradoxical respiration associated with lip and peripheral cyanosis. The abdomen was distended with
tenderness, without peristalsis. The rest of the examination was unremarkable. Abnormal laboratory findings included: white blood cells 20,280 mm$^3$ (neutrophils 91%), C-reactive protein 212 mg/l, glucose 1.28 g/l, blood urea nitrogen 1.24 g/dl, creatinine 3.37 mg/dl, potassium 5.4 mEq/l, creatine kinase (CK) 26,892 U/l, CK-MB 666 U/l, lactate dehydrogenase (LD) 7135 U/l, aspartate aminotransferase (AST) 1142 U/l, alanine aminotransferase (ALT) 880 U/l, amylase 2843 U/l, lipase 749 U/l, prothrombin time (PT INR 1.52, PTT RATIO 1.51). Blood gases showed acute metabolic acidosis with a blood pH of 6.79, bicarbonate 6.9 mEq/l, PaCO$_2$ 36 mmHg, bases excess $-26.2$. Electrocardiography showed sinus tachycardia. Electroencephalography showed signs of diffuse cerebral suffering. Left pleural fluid was present in chest X-ray. Abdominal X-ray and CT scan exhibited free subdiaphragmatic air and diffuse thickening of the gastric wall. The nasogastric tube was passed and drained conspicuous bloody-biliary material and gas. Resuscitation measures were started by intravenous hydration and correction of metabolic abnormalities to restore blood pressure and urine output. Thus the patient was sent to surgery. At laparotomy, a massive gastric necrosis, with evidence of a little perforation in the posterior wall, and conspicuous bloodstained peritoneal fluid were found. The gastric wall appeared thickened and crackling (Fig. 1). A total gastrectomy and a Y-Roux esophagojejunostomy was performed. In the histological exam the specimen presented massive dilatation and gangrene. The mucosa appeared thin and flat. Microscopy revealed full-thickness necrosis of the gastric wall without a complete involvement of the cardia. No evidence of vascular thrombosis was found. The peritoneal fluid exam was negative.

The post-operative course was stormy. The 1st post-operative day, after a cardiac arrest, the patient needed to be moved to Intensive Care Unit. The 10th postoperative day a new intervention was performed for intestinal occlusion due to volvulus of the ileum. The remainder postoperative period was characterized by a severe nutritional deficit, treated with artificial nutrition. Actually, at the 1-year follow-up, the patient presents normal oral feeding, with weight incremented by 20 kg. No breathing distress episodes occurred.

3. Discussion

Many patients affected by RS present oropharyngeal problems and breathing dysfunction. The physiological knowledges are poor but the underlying neurological defects is probably an immaturity of neurons regulating breathing mechanism and of the autonomic nervous system. The oropharyngeal motility has been extensively investigated in these patients by videofluoroscopy. The commonest abnormalities of swallowing function found are: diminished tongue mobility, reduced oropharyngeal clearance and laryngeal penetration during swallowing [5]. Abnormal breathing patterns described are hyperventilation, breath holding, apnoea and rapid and shallow breathing. Particularly during breath holding these patients swallow an excessive amount of air that may cause abdominal distention. As reported in literature, the consequent air bloat is more common in these patients than in other conditions associated with oropharyngeal problems and it is one of the secondary diagnostic features of this syndrome (Rett Syndrome Diagnostic Criteria Workgroup 1988) [6–8].

Moreover, these patients often present upper gastrointestinal dysfunction. In fact oesophageal dysmotility is characterized by absent primary and secondary waves, delayed emptying and atony, while gastric dysmotility is also characterized by diminished peristalsis and atony [5]. During air swallowing, the stomach stretches, creating significant tension. If the girl with RS is unable to burp or pass gas, the bowel wall may become thin and an extreme distention of the wall of the stomach may lead to rupture.

Another common characteristic described in RS is chronic constipation, due to multiple factors such as lack of physical activity, poor muscular tone, diet, drugs, inadequate fluid intake, scoliosis and pain and discomfort associated with elimination. Bowel movements occur less frequently and this leads to a large accumulation of stool at the sigmoid and rectum. The prolonged distention of the rectum and the sigmoid from this large amounts of stool cause loss of muscle tone and consequently the bowel becomes large and baggy [7].

In our case we think that a multifactorial etiology is involved. In fact, the patient presented hyperventilation with breath-holding, air swallowing, gastroesophageal dysmotility and one-week constipation, all typical symptoms reported in RS. Clinical, laboratory and histological findings excluded thrombosis, infections and the other common etiopathogenetic conditions found in literature.

Fig. 1. The stomach appears totally necrotic with massive dilatation.
In 1970 Edlich demonstrated that a massive gastric dilatation results in a decrease intramural blood flow when the intragastric pressure exceeds 30 cmH\textsubscript{2}O [9]. At this pressure regimen the consequent occlusion of the venous drainage due to the high intraluminal tension has been surely a highly significant factor in our case too [2]. Moreover we believe the abnormal gastric dilatation with compression of the inferior vena cava caused a decreased venous return, with consequent marked hypotension. Thus the associated splanchic vessel sequestration worsened tissue hypoxia and metabolic acidosis.

As reported in literature, patients in this clinical status are generally treated with the positioning of a gastrostomy that permits the air removal through gastrobutton [10]. A probable diagnostic delay, partially justified by the rarity of this complication, carried out to the several gastric ischemia. Total gastrectomy was the only treatment possible.

We believe that an early detection as well as consultation with a gastroenterologist or a surgeon are extremely important to avoid progression of the problem and to manage it as early as possible, thus preventing severe complications, as gastric necrosis. This might include the placement of a nasogastric tube or a gastrostomy to decompress the bowel and allow the gas to flow out.

Moreover during the first post-operative period the support of the artificial nutrition may help to reduce the weight loss and to guarantee a gradual resumption of the intestinal function. In this case, at 1-year follow-up, the girl did not present more breathing distress episodes and appeared more calm, with less episodes of anxiety.

Usually the irregular breathing patterns become less noticeable as these patients get older, but we retain that the accurate post-operative monitoring and the assiduous surveillance of the psychophysical status for a long period remain essential in RS.

References