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Combined Extrinsic and Intrinsic Duodenal Obstruction in a Down's Syndrome Child: A Diagnostic Challenge

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Abstract

Introduction: Down's syndrome is associated with various gastrointestinal disorders such as motility disorders, autoimmune phenomena, and structural abnormalities. Within these pathologies, incomplete forms of intestinal occlusion pose a diagnostic challenge due to their larval symptomatology.

Clinical Case: We present the case of a 2-year-old boy with Down's syndrome. He was referred to Gastroenterology for projectile vomiting from the start of complementary feeding. An abdominal ultrasound, opaque enema, and manometry with rectal biopsy without alterations were provided, as well as negative serology for Coeliac disease. On examination, there was marked distension of the upper abdomen, with palpation of the intestinal loops. In the oesophagogastroduodenal transit, a partial obstruction of the second portion with associated malrotation was observed, which was corrected surgically. The symptoms reappeared 15 months later finding a duodenal web in the third portion.

Discussion: Duodenal atresia affects 1 in 10,000 live births and may be associated with other congenital anomalies. Up to 30% have trisomy of chromosome 21. Duodenal web is an uncommon form, with incomplete obstructions presenting postprandial vomiting and distension of the upper hemiabdomen with visible peristalsis. The diagnostic test of choice is oesophagogastroduodenal transit and treatment requires surgical repair of the defect.

Conclusion: Structural digestive manifestations associated with Down's syndrome are a frequent cause of morbidity and mortality. Their management requires a high index of suspicion to indicate the necessary complementary tests, obtain an accurate diagnosis and establish early treatment.

Keywords: Down's syndrome; Duodenal atresia; Intestinal occlusion; Duodenal web; Oesophagogastroduodenal transit

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Introduction

Down's syndrome is a genetic disorder that occurs when abnormal cell division produces an extra copy of all or part of chromosome 21. This syndrome owes its name to Dr. Down who in 1866 first described the peculiar clinical features of a group of patients. Almost a century later, geneticists Jérôme Lejeune and Patricia Jacobs discovered the underlying genetic alteration [1].

It has an incidence of one in every 600 to 700 conceptions [2]. Only one risk factor has been demonstrated, maternal age (especially when the mother is over 35 years of age) and, very exceptionally, in 1% of cases, it is caused by parental inheritance. It occurs in all ethnic groups, in all countries and in all social classes. In our environment, this syndrome is the main cause of intellectual disability and is the most common human genetic disorder [1,2].

This syndrome is frequently associated with various systemic manifestations such as congenital heart disease in up to 50% of cases, acute myeloid leukemia, bone, hearing, visual, endocrinological and gastrointestinal disorders [3]. For this reason, interdisciplinary management is essential, as well as knowledge of these associations to establish a diagnosis of suspicion.

The gastrointestinal disorders associated with this syndrome include structural abnormalities, autoimmune phenomena, and motility disorders [1-3]. It can be a diagnostic challenge to determine

the type of underlying pathology due to the non-specific and latent clinical manifestations, but the nuances of the clinical history and physical examination will allow us to orient the case.

Clinical Case

We present a 2-year-old toddler who was referred to the gastroenterology department for vomiting [4]. He was the younger brother of a fraternity of 7 siblings of non-consanguineous parents, with a history of psychomotor retardation and learning difficulties in 4 of the siblings.

Pregnancy was monitored, with close follow-up due to the detection of polyhydramnios on ultrasound at 24 weeks. The baby was born prematurely at 34+5 weeks, without requiring resuscitation.

Examination of the newborn showed a high weight for gestational age (3100 gm, p95) with the rest of the anthropometry in accordance, a pansystolic IV/VI murmur in the left fourth intercostal space radiating to the apex, and a Down's phenotype. Chromosome trisomy was confirmed by karyotyping.

As background information, he was under follow-up in Cardiology for small muscular ventricular septal defect and mild sub pulmonary stenosis, as well as in Pneumology for recurrent bronchospasm. Vaccination was up to date.

Regarding the chronology of feeding, he had been exclusively breastfed for the first two months, with introduction of complementary feeding at five months, at which time he started vomiting.

The vomiting was late postprandial, not predominantly hourly, 1-2 per day, projective and with the content of food digested hours earlier. There were also episodes of abdominal distension which the parents clearly identified in the right hemiabdomen.

The parents provided a series of tests previously performed in another center, including negative serology for Coeliac disease, abdominal ultrasound without alterations and an opaque enema with redundant sigma without transition zones and stenosis. They also submitted a manometry with positive anal inhibitory reflex at high volumes, and a rectal biopsy showing ganglion cells in the mucosa, ruling out Hirschsprung's disease.

Physical examination revealed normal anthropometry, with a good general but poor nutritional condition, with soft muscle masses and preserved adipose panniculus. There was marked distension of the upper hemiabdomen and palpable bowel loops.

Considering the clinical history and the findings on examination, it was decided to request an oesophagogastroduodenal transit (Figure 1).

The images showed a stop of the barium column at the level of the 1st and 2nd portion of the duodenum, with reflux of papilla towards the gastric chamber. After 30 min, it began to empty. The duodenum was also found to be in an anomalous position, with the Treitz angle to the right of the midline. All of this was compatible with partial obstruction of the duodenum in the second portion with associated malrotation.

The patient underwent surgical correction, and the procedure revealed congenital Ladd's bands and intestinal malrotation. The surgical procedure consisted of evisceration, resection of the area of partial obstruction and Ladd's bands. The surgery also included an appendectomy with placement of small bowel loops in the right hemiabdomen and large bowel loops in the left hemiabdomen.

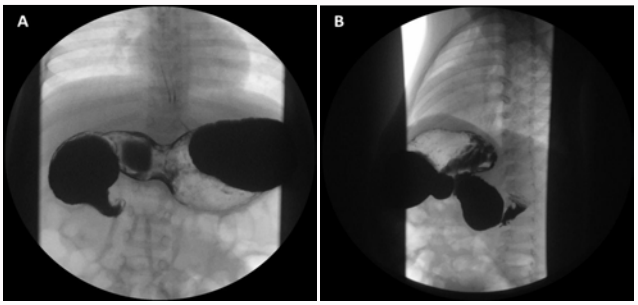

Figure 1: A) Anteroposterior radiological view showing a stop of the barium column between the 1st and 2nd portion of the duodenum, with reflux of papilla towards the gastric chamber. B) The same image can be seen in the lateral image.

He presented with sepsis in the immediate postoperative period, requiring admission to the intensive care unit, with a favorable outcome.

Fifteen months after the intervention, he consulted for persistent episodes of vomiting and abdominal distension, with imaging tests showing findings compatible with persistent duodenal obstruction. Surgery revealed a mobile duodenum with a dilated 1st and 2nd portion and an abrupt change in caliber in the 3rd portion, without extrinsic stenosis. The procedure consisted of a duodenotomy in the dilated portion, revealing the existence of a windsock web of the duodenum. A duodenoduodenostomy excluding the affected area was performed. There were no postoperative complications, and the patient remained asymptomatic from the gastrointestinal point of view.

Discussion

Duodenal obstruction is a rare cause of intestinal obstruction [5]. It can be due to extrinsic (annular pancreas, congenital flanges or malrotation) and intrinsic causes. Intrinsic obstruction may be due to atresia, stenosis, or duodenal web (total or partial), although intrinsic obstruction is commonly referred to as atresia [5,6].

Duodenal atresia is an embryopathy of the cranial intestine due to a failure of vacuolization and recanalization of the solid phase of duodenal embryogenesis that occurs from the 5th to the 10th week of gestation [7].

It affects 1 in 10,000 live newborns according to the series and may be associated with other congenital anomalies. More than 30% of cases are associated with Down's syndrome, about 40% have cardiac malformations and up to 70% are associated with intestinal malrotation [8].

Duodenal web is a rare cause of intestinal obstruction and its clinical presentation will depend on the degree of obstruction. In the prenatal period it may manifest as polyhydramnios in up to 20%, as in our case. In the postnatal period, symptoms may be early if the obstruction is severe or, more frequently, late, manifesting as growth retardation, post-prandial vomiting, and distension of the upper hemiabdomen with visible peristalsis [8,9].

Although ultrasound and abdominal X-ray can help in the diagnosis of suspicion, the gold standard is the oesophagogastroduodenal transit, which also makes it possible to identify the point of obstruction, to see whether it is total or partial and to rule out malrotation.

The usual treatment for this disease consists of surgical repair of the defect. This is the first case of combined extrinsic and intrinsic duodenal obstruction in a Down's syndrome patient described in the literature, highlighting the need for adequate surgical revision to rule out associated alterations. Endoscopic treatment could be an alternative, although the current evidence is limited [10].

Conclusion

Down's syndrome is often associated with various systemic manifestations, making interdisciplinary management essential. Gastrointestinal disorders associated with this syndrome include motility disorders, autoimmune phenomena, and structural abnormalities.

The structural digestive manifestations associated with Down's syndrome are a frequent cause of morbidity and mortality and, therefore, a diagnostic challenge.

Their management requires a high index of suspicion to indicate the necessary complementary tests, obtain an accurate diagnosis and establish early treatment.

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