Duodenal Atresia and Hirschsprung Disease in a Patient with Down Syndrome

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ABSTRACT

A two days-old newborn female patient with Down Syndrome was admitted to our hospital with complaint of vomiting. Physical examination was unremarkable except for the typical physical appearance of Down Syndrome. An abdominal radiography showed the double-bubble sign, characteristic for duodenal obstruction, and the patient was operated with prediagnosis of duodenal atresia. However, during the operation, Hirschsprung's disease was suspected and the diagnosis was confirmed by rectal biopsy. In this study, we described the case of duodenal atresia together with Hirschsprung's disease in a patient with Down Syndrome. Radiologists and pediatric surgeons should consider this issue for a correct diagnosis and treatment.

Key words: *Duodenal atresia; Hirschsprung's disease; Down Syndrome*

Down Sendromlu Bir Hastada Hirschsprung Hastalığı ve Duodenal Atrezi

Down sendromu olan iki günlük yenidoğan kız hasta kusma şikayeti ile hastanemize başvurdu. Fizik muayenede Down sendromu'nun tipik fiziksel görünümü dışında özellik yoktu. Abdominal radyografide, duodenal obstrüksiyon için karakteristik olan çift kabarcık işareti izlendi ve hasta duodenal atrezi ön tanısı ile ameliyat edildi. Ancak operasyon sırasında, Hirschsprung hastalığından şüphelenildi, kesin tanısı rektal biyopsi ile doğrulandı. Bu çalışmada, biz Down sendromlu bir hastada Hirschsprung hastalığı ile birlikte olan duodenal atrezi olgusunu tanımladık. Radyolog ve çocuk cerrahları doğru tanı ve tedavi için bu durumu göz önünde bulundurmalıdır.

Anahtar kelimeler: *Duodenal atrezi, Hirschsprung hastalığı, Down Sendromu*

INTRODUCTION

Down Syndrome (DS) is a common chromosomal anomaly resulting from trisomy 21 (1). The incidence is approximately 1 in 700-800 live births [2]. It is clinically diagnosed by its characteristic features including craniofacial dysmorphic physical appearance, mental retardation and systemic anomalies such as cardiac and gastrointestinal defects (2). The association of DS with anomalies of gastrointestinal system such as tracheoesophageal fistulas, duodenal atresia (DA) esophageal atresia, imperforate anus and Hirschsprung's disease (HD) is well established (1,3). The DA associated with HD in a patient with DS is rare presented in literature (4,5). This case report describes the DA associated with HD in a patient with DS.

CASE

A two days old, 2500 g weighing newborn female with DS was admitted to our hospital with complaint of vomiting. Physical examination was unremarkable with the exception of characteristic physical appearance of DS. An abdominal radiography detected the double-bubble sign, classical for duodenal obstruction (Figure 1), and the patient was operated with the prediagnosis of DA. In operation a duodenojejunostomia was performed because of the duodenal atresia. During the exploration, the colon 10 cm proximally to the peritoneal reflection seemed to be narrow and above this segment of colon approximately 10 cm segment was dilated and transverse colon was much more dilated. So on with the suspect of congenital megacolone, we performed a peroperative frozen section from the narrowed segment of distal colon. The diagnosis was suspicious and full-thickness biopsy from each three segments of colon then proximal loop colostomy was performed. The histopathological diagnosis revealed a ganglion negative specimen within the narrowed segment and that was compatible with HD (Figure 2). Since the thyroid hormone levels within the postoperative blood sample was compatible with hypotiroidia, medical treatment was attended. Patient's chromosomal analyses, which revealed that the karyotype was 47 XX, were normal except trisomia 21. The renal ultrasonography and an echocardiography did not determine any abnormalities, and the patient was discharged from the hospital on the postoperative twelfth day. The case prospectively after the term of one year attaining to 8500 g of weight was admitted to our emergency service with severe enteritis. The patient was hospitalized to the intensive care unit with the diagnosis of dehydratation and sepsis due to enteritis. On the third day of treatment the patient died as a result of severe septicemia.

DISCUSSION

Knowledge of the embryologic development is necessary to comprehend the gastrointestinal system anomalies such as DA and HD. DA is believed to be related to errors of recanalisation of the solid cord stage of developing embryonic foregut in early gestation (6,7). HD is caused by the failure of colonic ganglion cells to migrate cephalocaudally through the neural crest in gestation (8). It is a developmental disorder of the enteric nervous system and is characterized by the congenital absence of ganglion cells in the myenteric and submucosal plexuses of the distal gut (8,9). Duodenal atresia is a common cause of congenital, intestinal obstruction. The incidence has been estimated at 1 in 6.000 to 1 in 10.000 births (6). The atretic segment is most often the second part of the duodenum, distal to the duodenal papilla (10). HD is an intestinal innervation disorder that causes development of functional intestinal obstruction. It occurs 1 in 5000 births (8,9). The aganglionic segment usually begins at the anus and extends proximally (8). Short-segment disease is most common and is restricted to the internal sphincter and rectosigmoid region of the colon. Long-segment disease is less common and can affect the total colon (8,9). DA can be associated with many congenital anomalies such as esophageal atresia, intestinal malrotation, imperforate anus, Meckel's diverticulum, cardiac and renal anomalies. There is also a rare association with anomalies of biliary tract (11). HD can be associated with cardiovascular, urologic, neurologic and gastrointestinal abnormalities such as ventricular septal defect, bladder diverticulum, renal agenesis, cryptorchidism, neuroblastomas, hydrocephalus, imperforate anus, Meckel's diverticulum. It has been also linked pigment defects and primary alveolar hypoventilation (8). Approximately 30% of patients with DA (9) and 10% of patients with HD (8) have DS (trisomy 21). Because of the complications of DA and HD, early diagnosis and treatment should be of great value. Imaging can help diagnosing DA and HD. In DA, the double bubble sign is a finding detected on plain abdominal radiographs of newborns and infants. The finding shows two airfilled structures due to stomach and duodenum in the upper

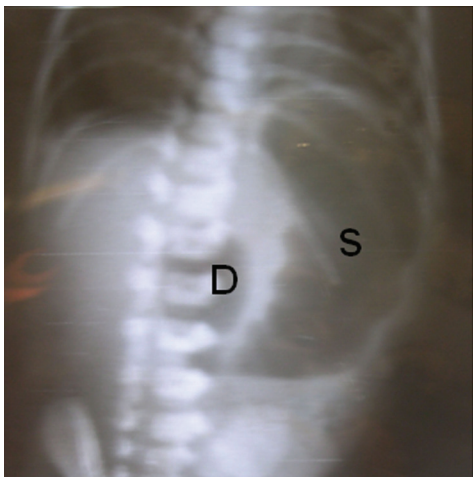

Figure 1. Plain radiography of abdomen shows the classic double-bubble sign of duodenal atresia. S: Stomach, D: Proximal duedonum.

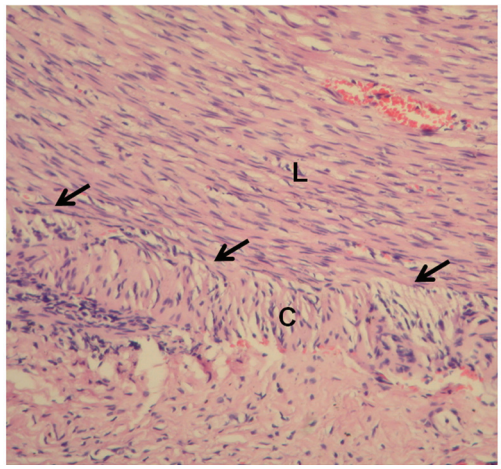

Figure 2. Microscopic section shows ganglion cells negative (arrows) specimen between the longitudinal (L) and circular (C) muscle layers of the bowel.

abdomen. It also shows the paucity or absence of distal bowel gas (12). The ultrasonography can be also used to demonstrate the double bubble sign. However, imaging may be difficult and time consuming especially when there is a gas distention in the abdomen. In addition, it is an operator-dependent examination. If the classic appearance of a double bubble is determined on plain abdominal radiographs in the neonate, additional radiologic investigation is unnecessary, and the surgeon is considered to plan for surgery (12). The diagnosis of HD is based on the medical history, the plain abdominal radiograph, the contrast enema radiography results, and the full-thickness rectal biopsy findings. A plain abdominal radiograph may show a dilated small bowel or proximal colon (8). The pathognomonic finding of HD on contrast enema is a transition zone between the normal and aganglionic bowel. Typical operative findings of HD are of apparently normal distal rectum and a massively dilated proximal colon (9). The diagnosis is confirmed with the full-thickness rectal biopsy, which should show the absence of ganglion cells (8). In this case, the DA was determined on plain abdominal radiograph, and the patient was operated. However, HD was determined during operation, and the diagnosis was confirmed by rectal biopsy.

In conclusion, association of DA and HD in a patient with DS is feasible; therefore radiologists and pediatric surgeons should regard this issue for a correct diagnosis and treatment.

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