CASE REPORT

Association of Duodenal Atresia, Malrotation, Ventricular Septal Defect, Endocardial Cushion Defect, Atrial Septal Defect, and Patent Ductus Arteriosus in a Down Syndrome Patient

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ABSTRACT

Duodenal atresia is the frequent cause of neonatal intestinal obstruction. The association between duodenal atresia, intestinal malrotation, cardiac anomalies, and Down syndrome is infrequently reported. We present a prenatally suspected case of duodenal atresia which was associated with malrotation and cardiac anomaly in a patient of Down syndrome.

Key words: Cardiac anomaly, down's syndrome, duodenal atresia

INTRODUCTION

It is seen in two ways intrinsic (atresia, stenosis, and web) or extrinsic (malrotation, Ladd's bands, annular pancreas, and duplication) as congenital intestinal obstructions.[1] Duodenal atresia associated with the defect and recanalization of the distal duodenum which develops from the midgut with the proximal duodenum that develops from the midgut occurs. It is seen in 1–2.8% of 10,000 live births during neonatal period, the most common cause of small bowel obstruction.[2-4] Duodenal atresia is seen in 20–30% of newborns with Down syndrome. It can be detected by ultrasonography performed during prenatal period. Duodenal atresia may be accompanied by polyhydramnios in the prenatal period as the amniotic fluid prevents absorption of the fluid from the intestines.[5] 30–50% of patients with duodenal atresia are isolated and the rest may be accompanied by cardiac, vertebral, gastrointestinal, or renal anomalies. Trisomy 21 may be present in 20–30% of cases.[6] We present a prenatally suspected case of duodenal atresia which was associated with malrotation and cardiac anomaly in a patient of Down syndrome. The importance of prenatal diagnosis and postpartum follow-up was emphasized with this case being presented.

CASE REPORT

A 1-day-old boy (weight 2.5 kg), born through cesarean section at 37+5, was evaluated for prenatal ultrasound findings of double bubble sign and polyhydramnios. APGAR score was 9 in the 1st min and 10 in the 5th min. Physical examination revealed Mongol facial appearance, upward slanting palpebral fissures, hypothyroidism, nasal root compression, brachycephaly, short and thick neck, clinodactyly on the right and left hand fingers, simian line on the hands, sandal gap deformity on the foots, and 2/6 systolic murmur [Figure 1]. The patient had typical features of Down syndrome and was already proven on prenatal testing. He with feeding difficulty and vomiting was transferred to intensive care unit for stabilization. Abdominal radiograph showed double bubble sign [Figure 2]. Barium fluoroscopy was taken, double bubble sign confirmed [Figure 3]. Nasogastric tube was placed. Echocardiography revealed perimembranous ventricular septal defect (VSD), atrial septal defect (ASD), endocardial cushion defect, and patent ductus arteriosus (PDA). The patient was hemodynamically stable. Duodenoduodenostomy and Ladd's procedure were performed. A trans-anastomotic tube and abdominal drain

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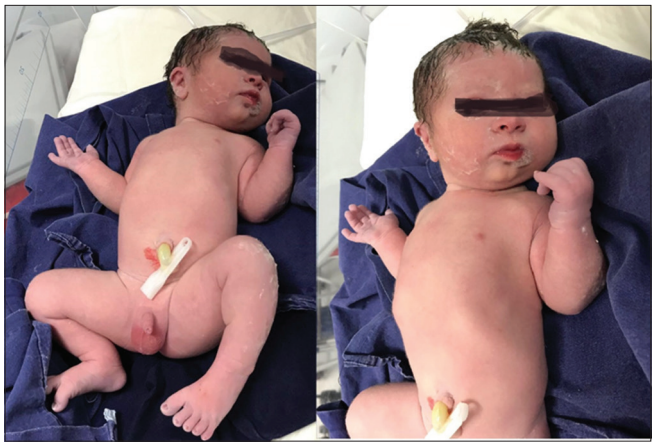

Figure 1: Our patient with Down syndrome

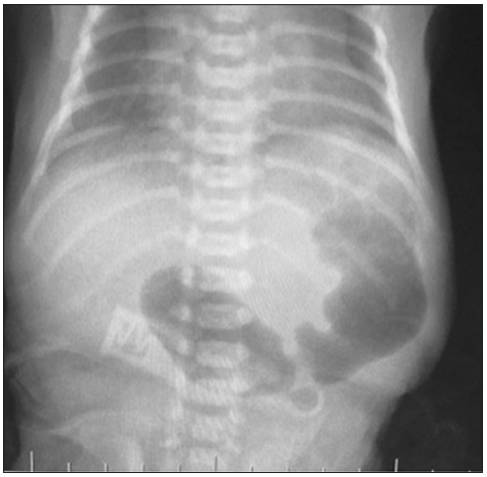

Figure 2: Abdominal radiograph showed double bubble sign

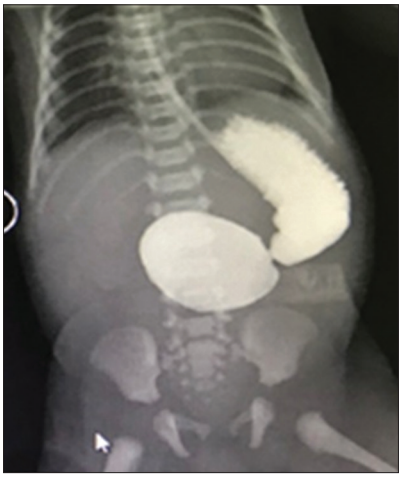

Figure 3: Barium fluoroscopy showed double bubble sign

were placed. The patient started orally on the 10th post-operative day after upper gastrointestinal contrast study which was reported as normal. The postnatal 6th month was planned surgery for cardiac anomaly. Genetic analysis resulted in 47, XY, + 21 (Trisomy 21).

DISCUSSION

It was first described by Calder in 1733. The mortality rates of duodenal atresia were 86–90% in the past and have decreased to 4% by surgical methods. The presence/absence of associated anomalies has an important impact on prognosis. The mortality is mainly due to associated complex cardiac anomalies.[2-4] The association of duodenal atresia with Down syndrome, and vertebral, anorectal, cardiac, transesophageal, renal, and extremity anomalies syndrome has been described.[7]

Duodenal atresia is presented with vomiting and feeding difficulty in the early stages of life. It usually develops during the first 24–38 h period after initial feeding and progresses progressively if not treated. Abdominal distention occurs in patients without intestinal motility.[8] Our patient had feeding difficulties and vomiting.

Prenatal ultrasonography may show signs of Down syndrome and a double bubble. The initial postnatal radiographic evaluation for diagnosing duodenal atresia is a plain abdominal X-ray. A double bubble sign on an abdominal X-ray is a reliable indicator of duodenal atresia. Other causes of intestinal obstruction may simulate a double bubble sign. Annular pancreas is the second most common cause of duodenal atresia. Jejunal or more distal obstruction may have dilation more distally or more than two bubbles may be present. The radiographic appearance of the double bubble sign should prompt immediate surgical consultation. For cases of suspected duodenal atresia not identified antenatally, barium fluoroscopy can be used to assess the gastrointestinal tract. The main purpose of the upper gastrointestinal series is to differentiate between duodenal atresia and midgut volvulus; an important distinction, because midgut volvulus requires emergency surgery, whereas duodenal atresia can be managed on an urgent basis.[8] Our patient had a double bubble sign. For differential diagnosis, barium fluoroscopy was performed and the diagnosis was confirmed.

Factors determining prognosis in babies with duodenal atresia are gestational week of birth, presence of congenital anomalies, and delay in diagnosis. Delay in diagnosis; vomiting, aspiration pneumonitis, electrolyte imbalance, dehydration, and stomach perforation can cause death. [9] The treatment of congenital duodenal atresia is to ensure the continuity of the gastrointestinal passage. Duodenoduodenostomy is the most preferred surgical technique for this purpose.[10]

CONCLUSION

Early diagnosis and appropriate surgical treatment are important in patients with duodenal atresia. Prenatal ultrasonography may show signs of Down syndrome and a double bubble. We believe that delivery of prenatally diagnosed patients in centers with pediatric surgery will decrease mortality. As the association of duodenal atresia, malrotation, VSD, endocardial cushion defect, ASD, and PDA in a Down syndrome patient is rare in the literature, it was intended to be emphasized.

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