Intermittent volvulus with obstruction due to a Meckel's Diverticulum and band presenting as feeding intolerance in a neonate with Trisomy 13

Suresh Kotinatot[1]*, Shiva Shankar[1], Muhammad Ba'Ath[2], Ahmed Elajab[1] and Ajay Prashanth Dsouza[3]

[1]Department of Neonatology, Al Jalila Children's Hospital Dubai
[2]Department of Pediatric Surgery, Al Jalila Children's Hospital Dubai
[3]Department of Radiology, Al Jalila Children's Hospital Dubai

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*Corresponding author: drsureshk06@gmail.com

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Abstract

The most common malformations associated with Trisomy 13 are median facial or central nervous system defects;and a majority of those with the condition suffer from congenital heart defects.Meckel diverticulumexists in 1-2% of the general population, may be with much higher frequency in those with trisomy 13. It is however rarely symptomatic and such reported cases are extremely rare. We here describe neonate with feeding intolerance and bilious aspirates as a result of Meckel's Diverticulum and band causing intermittent volvulus with obstruction.

Keywords: *Trisomy 13, Bilious aspirates, Meckel's Diverticulum, Volvulus*

Introduction

Trisomy 13 or PatauSyndrome (PS)is an extremely rare but serious aneuploidy that occurs in 1 in 12,000 births, with a 91.4% mortality rate within the first year afterbirth. It results from either nondisjunction or Robertsonian translocation. [1] The most common malformations are median facial or central nervous system defects; and the majority of those with the condition suffer from congenital heart defects. [1] Although a Meckel's diverticulum (MD) is normally present in approximately 2% of the general population, its clinical presentation in the neonate is rare. [2]MD occurs at a much higher frequency of 25% amongpatientswithtrisomy13; however, symptomatic MD in patients with PS is extremely rare. [4]Only two casesdescribed

in the literaturefor infants with PS and symptomatic MD.Here, we report a case of a full-term neonate with trisomy 13 and Meckel's diverticulum who presented with bilious aspiratesand intermittent volvulus due to a MD with band.

Case presentation

A term male infant (3671 g, 41+2 weeks of gestation), delivered by normal vaginal delivery to 27 years' old prima-gravida mother with APGAR scores of 6 and 8 at 1 and 5 minutes; respectively. The pregnancy was normal, with normal prenatal serological screening results and normal antenatal ultrasound done in second trimester. There is history of second degree consanguinity.The newborn was noticed to have multiple congenital anomalieson physical examination: complete bilateral cleft lip/palate, low set ears, microphallus and bilateral undescended testes. Further evaluation after admission to NICU revealed vermian hypoplasia and posterior fossa cyst on screening cranial ultrasound. Echocardiography showed moderate PDA, small ASD, and Bicuspid aortic valve.The newborn was intubated and mechanically ventilated in view of respiratory distress and increased work of breathing.The abdomen was soft on palpation, appeared not-distended with normal passage of meconium. Feeds were initiated at 5 ml every 3 hours on day 2 of life. Feeds were tolerated well for 2 days but on the third day greenish aspirates were noted. The abdomen remained soft and non-distended. An upper GI contrast study was done whichshowed the duodenojejunal (DJ) flexure to be in a normal position but the upper bowel loops had corkscrew appearance concerning for volvulus. [Figure 1 and 2]

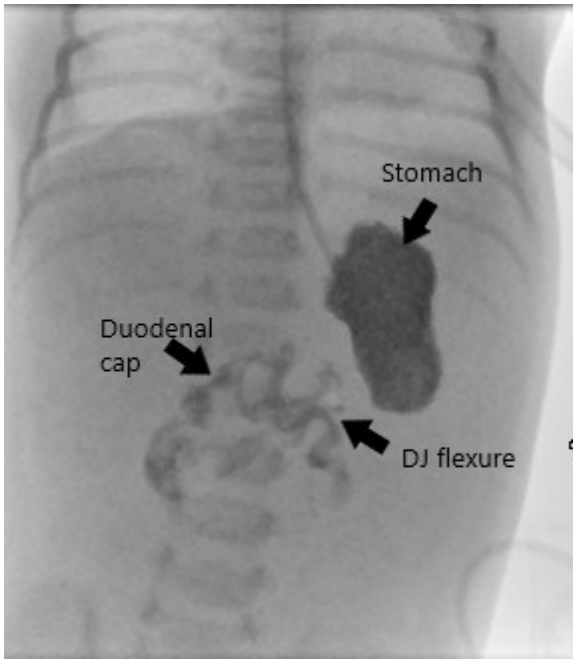

Figure 1: Fluoroscopic image of the upper GI contrast examination shows normal location of the stomach, duodenal cap and DJ flexure, to the left side of the spine and the same level as the duodenal cap.

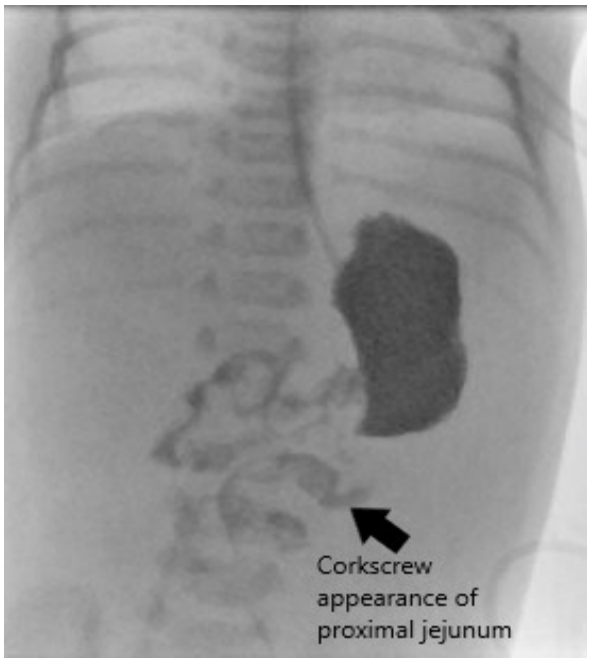

Figure 2: Fluoroscopic image of the upper GI contrast examination obtained distal to DJ flexure shows coiling of the proximal jejunal loop giving a corkscrew appearance concerning for volvulus.

Upon exploratory laparotomy, MD was found with peritoneal band extending from base of diverticulum to umbilicus likely causing intermittent volvulus and intestinal obstruction.

Small cyst like structure was noted in the middle of the band. No caliber change of bowel was noted at that level. The MD was resected and the bowel underwent primary anastomosis. Feeds were started on 4thpost-operativeday and increased quickly to full feeds. Karyotyping revealed Trisomy 13, 47, XY+13. Subsequently the child kept tolerating feeds and went on to have an uneventful laparoscopic assisted gastrostomy insertion. Pathology of the resected segment confirmed vitello-intestinal remnant on a small bowel segment in keeping with intra-operative findings.When correlated with the history of PS and the clinical presentation, the contrast examination findings led us to conclude that the cause of the bowel obstruction was likely due to symptomatic MD.

Discussion

Trisomy 13 or Patau Syndrome (PS) is one of the most common autosomal trisomies, and it is accompanied by anomalies of many organs.The relation of trisomy 13 to a clinical syndrome was first recognized by Patau et al in 1960. [3] Median survival time for patients with trisomy 13 is between 7 and 10 days and it is reported that between 86% and 91% of live-born patients with Patau syndrome do not survive beyond 1 year of life. Survival beyond the first year has been associated with mosaicism. The 19-year-old patient is the oldest known living person with regular trisomy 13.[17]

Meckel's diverticulum (MD) results from the formation of a true diverticulum in the small intestine as a result of an incomplete obliteration of the vitelline duct. Classically, it is often defined by the rule of two's: 1) occurs in approximately 2% of the population with a male-to-female ratio of 2:1, 2) Located within two feet from the ileo-cecal valve, 3) occurs before the age of 2 years, and 4) two inches in length; although, the size of a MD varies in practice. [18,19]. Although MD is the most common congenital anomaly of the gastrointestinal tract, its symptomatic manifestation in the neonatal period is rare. [10] Common presentations of neonatal Meckel’s diverticulum that have been reported in the literature include bowel obstruction by inflammation, perforation, intussusception, segmental ileal dilatation, and ileal volvulus. [12-16]

Symptomatic MD in PS is extremely rare and only few detailed reports are available in literature in spite of more than normal MD association as compared to normal neonates. [4] The coexistence of trisomy 13 and symptomatic Meckel diverticulum is reported in only 2 cases [4, 11] based on our literature search, although there are scattered case reports of trisomy 13 autopsies. In our case, MD was discovered at the time of laparotomy for suspected

volvulus with feed intolerance and bilious aspirates. MD has been diagnosed and reported earlier both during exploratory laparotomy for suspected volvulus/malrotation and more so as an incidental finding in postmortem of Trisomy 13 patients. [5-11]

"A neonate presenting with greenish aspirate or biliary emesis should be suspected for malrotation and volvulus unless proven otherwise. It warrants intensive care admission for nil per oral, parental intra-venous fluids, metabolic acidosis/alkalosis with electrolytes correction, urgent paediatric surgery consult, upper GI contrast study, abdomen USG with doppler as part of work-up." Volvulus on delayed images in the absence of features of malrotation of the duodenum based on the usual location of DJ flexure, is an unexpected finding. Usually, intestinal volvulus is associated with malrotation. The traditional teaching has been that the demonstration of a typical DJ flexure and the proximal jejunum's expected location can rule out the diagnosis. Considering the incidence of MD in general population and rarity of symptomatic presentation, it would not be cost appropriate to recommend Isotope scan routinely in all Trismoy 13. However the idea of considering the isotope scan for any GI symptoms in babies with Trisomy 13 is thought provoking. We think that even if it is negative (known to have 30% false negative rate) in a baby with features of volvulus/malrotation the need for urgent surgical exploration and correction will still be present. Although one can consider Isotope scan it may not help in preventing surgical exploration with abdominal features of volvulus/malrotation.

In our case, a significant point of learning is reflected in the review of the upper GI contrast examination. The radiologist performing the upper GI contrast study should look for the proximal small bowel loops for their location, morphology and contrast emptying, even though a clear demonstration of the normal DJ flexure is made. Otherwise, volvulus without malrotation can be missed.

The approach to treatment of MD depends on whether it is discovered incidentally or as a result of symptoms. The appropriate management of incidentally discovered MD has been controversial, and a physician may choose either to surgically resect or not to resect the incidentally discovered MD. In contrast, a case of MD resulting in symptoms must be treated by surgical resection as in our case. As most cases of MD in Trisomy 13 neonates have only been discovered on autopsy, the exact magnitude of symptomatic MD among newborns with Trisomy 13 is not yet determined.

Conclusion

Preoperative diagnosis of a symptomatic MD in a neonate is often difficult, so it is necessary to maintain a high index of suspicion aided by delayed images of the contrast study as an early surgical approach is essential to achieve a successful outcome.The co-occurrence of PS and symptomatic MD suggests possible association. MD should be considered in any child with PS that presents with bowel obstruction, especially intermittent. Standard surgical treatment of MD resection and primary bowel anastomosis is safe and effective.

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