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A Case Report: Colonic Atresia in a Newborn with Presumed Rubinstein-Taybi Syndrome

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Abstract: Colonic atresia is an uncommon cause of intestional obstruction. There is a scarcity of cases of colonic atresia described in the literature, and the pathogenesis of the disease remains unknown. Although the clinical presentation of patients with colonic atresia seldom varies, reported co-occurring anomalies vary widely; almost half of the cases involve other congenital defects. We report a case of colonic atresia that appears to have co-occurred with Rubinstein-Taybi syndrome, another rare congenital disease. Based on a literature search, we believe that this might be the first reported case of co-occurrence of these two rare anomalies.

Keywords: *colonic atresia, Rubinstein-Taybi syndrome, barium enema, bowel*

Key Points

- Colonic atresia is a rare intestinal atresia that typically presents with abdominal distension, failure to pass meconium after birth, and bilious emesis.
- Diagnosis of colonic atresia might be confirmed by examination with barium enema.
- Colonic atresia can co-occur with other intestinal atresias, gastroschisis, atrial septal defect, anorectal malformations, Hirschsprung disease, colonic duplication of the distal colon and, as was found in this case, with Rubinstein-Taybi syndrome.
- Patients with colonic atresia should be systemically evaluated for the presence of the co-occurring anomalies.

Abbreviations

CA: colonic atresia
RTS: Rubinstein-Taybi syndrome
MRI: magnetic resonance imaging

Introduction

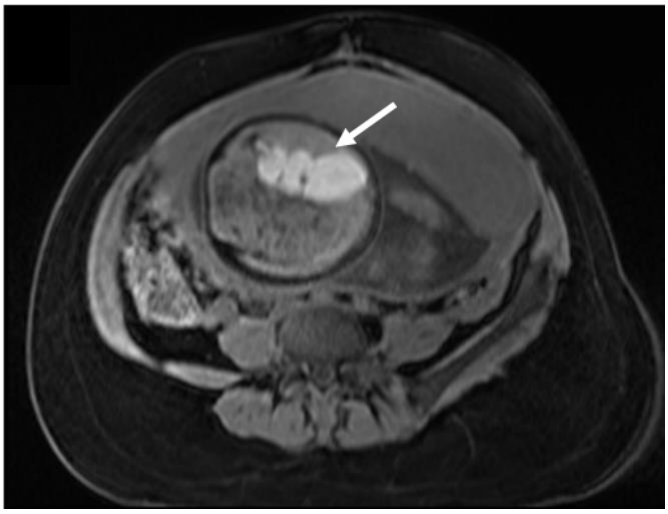
Colonic atresia (CA) is considered a rare form of intestinal atresia. The National Birth Defects Prevention Network reports the prevalence of rectal and large intestine atresias and/or stenoses at 4.2 per every 10 000 live births.[1] The clinical presentation of patients with CA typically includes distension of the abdomen, failure to pass meconium within 24 to 48 hours after birth, and bilious emesis.[2-4] In a review of 224 cases of CA, Etensel et al[5] found that the right side of the colon was more frequently affected by CA than the left side of the colon, and the transverse colon and the ascending colon were the most common sites of the disease.

We report a case of CA in a newborn with multiple congenital anomalies and clinically suspected Rubinstein-Taybi syndrome (RTS). Although the results of molecular genetic testing for identification of a heterozygous pathogenic variant in the *CREBBP* or the *EP300* genes were negative, clinical and radiologic findings were strongly suggestive of RTS, according to the patient's geneticist. Rubinstein-Taybi syndrome has been linked to genetic mutations in the *CREBBP* and the *EP300* genes, members of the histone acetyltransferases that function as

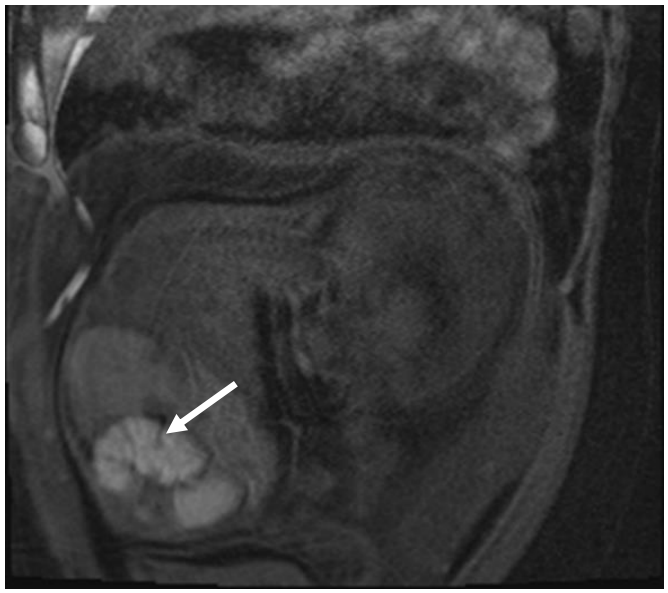
transcriptional coactivators.[6,7] However, genetic testing with confirmatory results were reported in only 55% to 78% of suspected cases, and the syndrome is mostly diagnosed by clinicians.[6,7] Rubinstein-Taybi syndrome, recognized as a syndrome in 1963 by Rubinstein and Taybi, has an estimated incidence of 1 in 100 000 to 125 000 live births and is characterized by broad and short first digits of the extremities, anomalies of the face and the skull, high-arched palate, growth retardation, and cognitive disabilities.[6,7] In a search of literature, we have found no reported cases of RTS that co-occurred with CA.

Figure 1. Fetal Magnetic Resonance Imaging (MRI) at Approximately 37 Weeks of Gestation.

A T1-weighted MRI, axial view



B T1-weighted MRI, saggital view

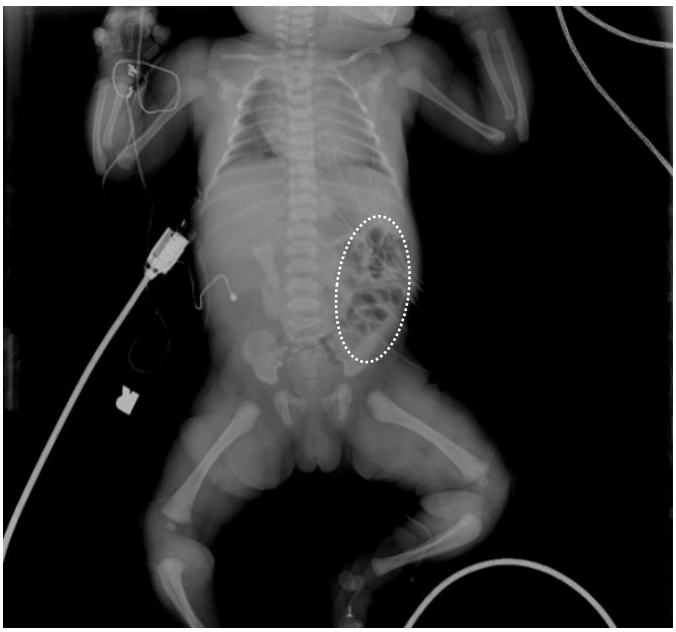


(A, B) T1-weighted fat saturated volumetric interpolated breath-hold examination (VIBE) MR images show axial and sagittal views of the fetus with a dilated ascending colon and cecum (A and B, arrows), as evidenced by the presence of haustra and T1-hyperintense meconium.

Case Presentation

The patient was born 39 weeks 0 days of gestation to a gravida 3, para 1, aborta 2 mother via scheduled caesarian delivery because of the complications associated with the multiple anomalies discovered in the antenatal period as well as the breech position in utero. The neonate's birth weight was 2 906 g. Apgar scores were not recorded. The mother's pregnancy was complicated by fetal anomalies, lack of immunity to measles, the breech presentation of the fetus, and group B streptococcal bacteriuria.

Figure 2. Radiography of the Chest and the Abdomen Performed Approximately One Hour after Caesarian Delivery at 39 Weeks 0 days of Gestation in an Infant with the Symptoms of Intestinal Obstruction.



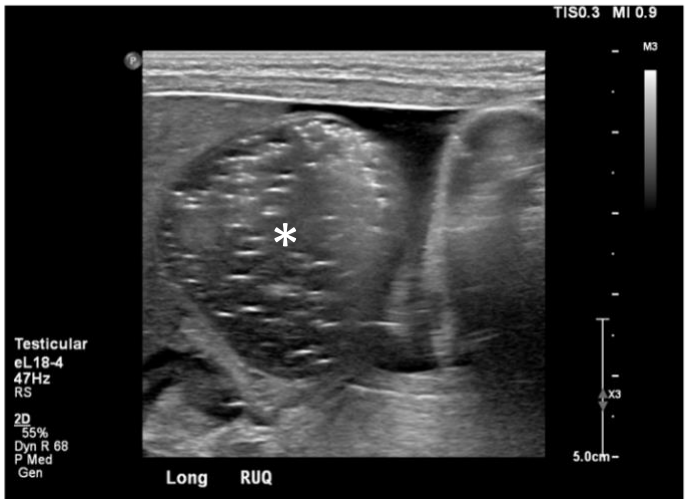
Anteroposterior-view image shows nondilated small bowel (dotted oval) deviated to the left side of the abdomen, suggesting the presence of intra-abdominal mass.

The first documentation of multiple fetal anomalies was made on obstetric ultrasonography at approximately 29 weeks of gestation. The images of this examination are not available. According to the electronic medical record, the anomalies included ventriculomegaly, absent corpus callosum, colpocephaly, a cystic lesion in the abdomen, and a left-sided stomach. Based on these findings, Trisomy 18 was suspected but subsequently ruled out by the result of genetic testing. Fetal magnetic resonance imaging (MRI) at approximately 37 weeks of gestation confirmed agenesis of the corpus callosum and revealed no abnormalities of the urogenital system or the abdominal wall, but did show a dilated ascending colon and cecum filled with meconium, as suggested by a notable T1-hyperintense signal throughout the lesion (Figure 1).

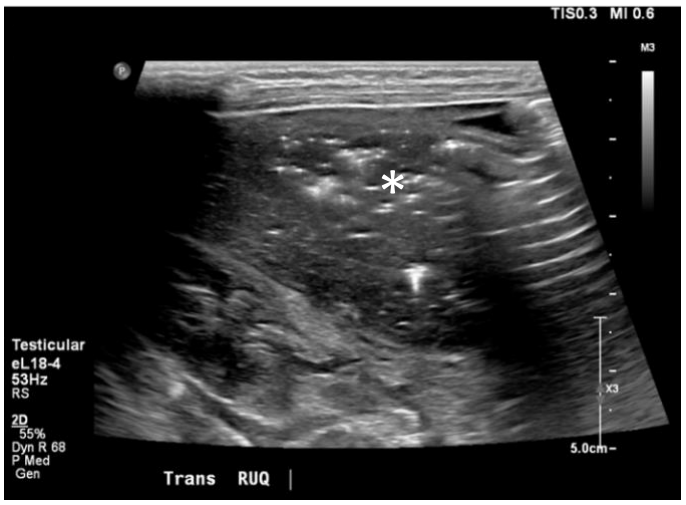
Findings from physical examination revealed a high-pitched crying infant with ectopic tissue protruding from the right orbit, bilateral cataracts, a high-arched palate, low-set ears, a systolic murmur, and rocker-bottom feet. Of note, the patient had a normal, patent anal canal. The results of initial point-of-care laboratory analysis showed hypoglycemia, 47 mg/dL (reference range, 65-99 mg/dL) and elevated total creatine phosphokinase, 339 U/L (reference range, 38-282 U/L). During the first 24 hours postnatally, the infant exhibited intermittent oxygen desaturations, poor sucking reflex, inability to

Figure 3. Ultrasonography of the Right Upper Quadrant of the Abdomen in a Newborn with the Symptoms of Intestinal Obstruction.

A Longitudinal view

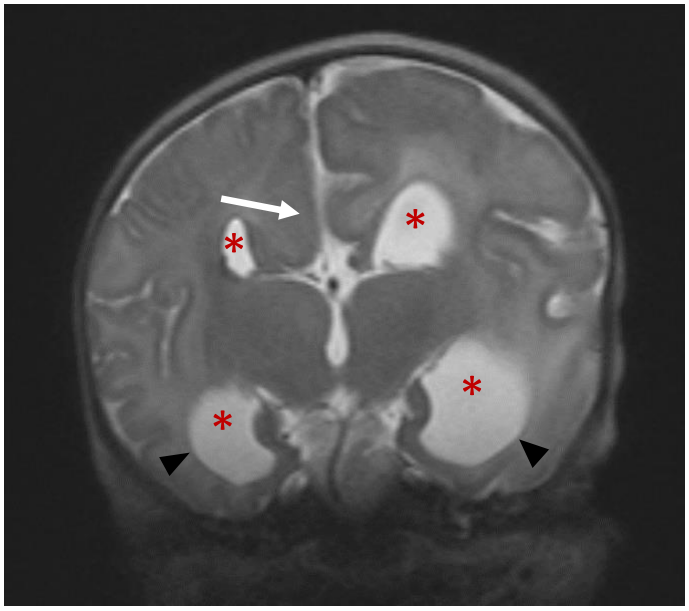


B Transverse view



(A, B) Longitudinal view and transverse view images show the dilated bowel (A and B, asterisks) filled with debris and debris-associated marked hyperechogenic foci, suggestive of the presence of meconium.

Figure 4. Magnetic Resonance Imaging (MRI) of the Brain Performed Approximately Nine Hours after Caesarian delivery at 39 Weeks 0 days of Gestation in an Infant with the Symptoms of Intestinal Obstruction and Clinically Suspected Rubinstein-Taybi Syndrome.



Half-Fourier acquisition single-shot turbo-spin echo (HASTE) coronal T2-weighted image shows a partial view of the complete agenesis of the corpus callosum (arrow) and a partial view of the marked ventriculomegaly (asterisks) with colpocephaly (arrowheads).

tolerate oral feeding, no passage of stool, and progressive abdominal distention. A double-lumen catheter (Replogle tube) that was placed within 24 hours had persistently minimal output.

An initial abdominal radiograph revealed aerated, nondilated small bowel and mass effect displacing the bowel to the left side of the abdomen (Figure 2). Follow-up sonography showed marked distention of the meconium-filled cecum and ascending colon (Figure 3). A postnatal MRI showed agenesis of corpus callosum and colpocephaly (Figure 4), which were previously detected on obstetric ultrasonography. A barium enema examination showed marked dilation of both the small bowel and the ascending colon (Figure 5A). An attempt to catheterize the rectum met significant resistance. Administration of contrast revealed a blind-ending rectosigmoid microcolon (Figure 5B, 5C), which was diagnostic of colonic atresia (CA).

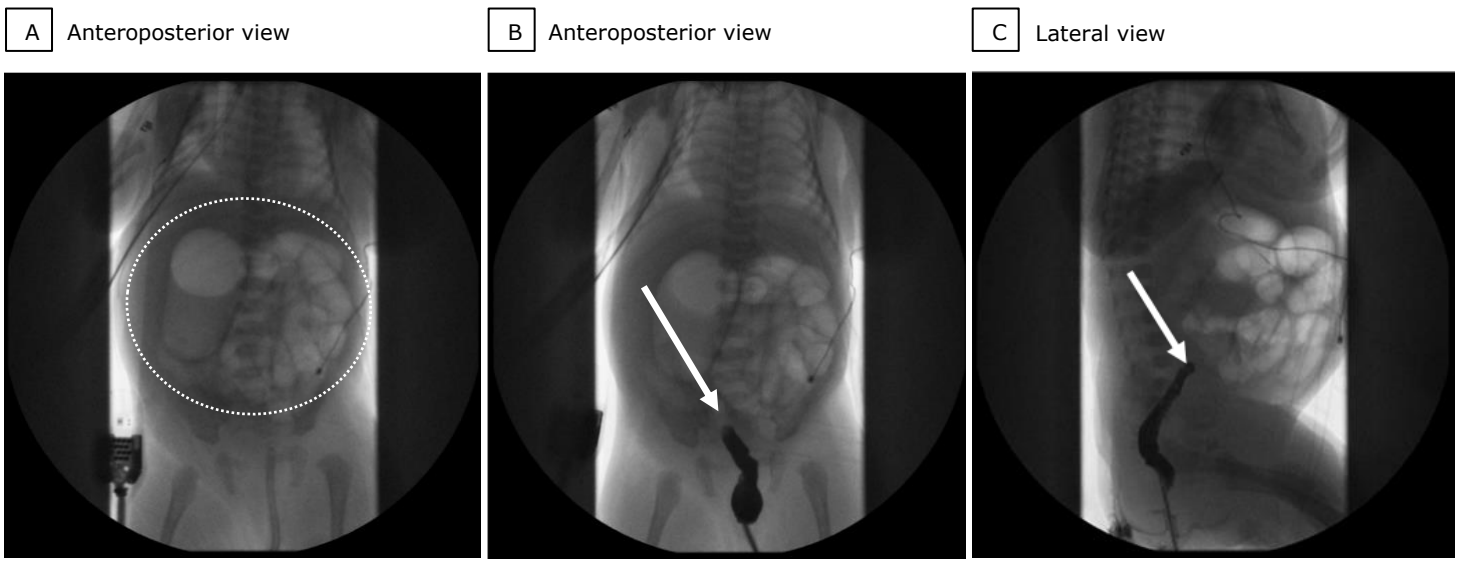
At two days of age, the patient underwent colostomy. Echocardiography later revealed a patent ductus arteriosus and secundum atrial septal defect, both with left-to-right shunting. Electroencephalography showed focal seizure activity.

Discussion

Colonic atresias (CA) are categorized as one of four main types, based on the structure and the shape of defects. Type I CA is characterized by the presence of a mucosal membrane in an otherwise normal colon and with intact mesentery; type II CA is identified by a fibrous-band connection between two atretic ends of the bowel; type III CA is subdivided into the subtypes IIIA and IIIB based on the shape of a mesenteric defect, the "V" shape and the "apple-peel" shape, respectively; and finally, type IV is used to describe CA that occur in multiples.[2,5] Unfortunately, in our case, the categorization of the type of CA was not established.

Etensel et al[5] found that almost half of CA cases were associated with other congenital defects. Most commonly, these congenital defects include other forms of intestinal atresia, abdominal wall defects, and mesenteric abnormalities.[1,2,4,5,8] However, to our knowledge, there were no reports of CA cases associated with Rubinstein-Taybi syndrome (RTS), although there have been reports of malrotation[9] and volvulus[10] co-occurring with RTS. Notably, our patient also presented with

Figure 5. Barium Enema Study Performed 22 Hours Postnatally in an Infant with the Symptoms of Intestinal Obstruction and Presumed Rubinstein-Taybi Syndrome



(A) Anteroposterior-view image obtained just prior to barium administration shows high-grade bowel obstruction with dilated loops of bowel throughout the abdomen (A, dotted oval). (B) Postcontrast anteroposterior-view image shows contrast opacifying a blind-ended rectosigmoid microcolon (B, arrow). (C) Postcontrast lateral-view image shows contrast opacifying the rectosigmoid microcolon noncommunicating with the rest of the colon (C, arrow).

complete agenesis of the corpus callosum. As reported by Ajmone et al,[11] callosal dysmorphism is the most common (73.6%) brain abnormality in patients with confirmed RTS.

Atresia of the colon has been reported as "the most favorable type of congenital intestinal atresia with respect to survival."[3] The mortality rate associated with CA has been reported[5] at 25.7%. Early detection of CA is crucial for survival; mortality rates significantly increase after 72 hours without surgical intervention.[5]

Radiologic evidence of colonic obstruction is imperative for early diagnosis of CA. Abdominal radiography performed in patients with CA often reveals a dilated intestine with air-fluid levels.[2,3] Approximately two-thirds of radiographs obtained with barium enema show a distal microcolon.[2] Barium enema examination is recommended as a part of the initial imaging studies as it can help define location of CA and differentiate CA from other lower bowel obstructive diseases, such as Hirschsprung disease or meconium plug.[2,3] T1-weighted MRI may also be a useful adjunct diagnostic tool for characterization of cystic bowel lesions especially those filled with meconium[12].

Louw,[8] based on retrospective analysis of cases and experiments with animals, suggested that intrauterine mesenteric vascular accidents during the late stage of fetal development were likely a cause of most colonic atresias. Other authors suggest that the reasons for these defects might be a failure of recanalization at the solid stage of intestinal development,[2,4,8] familial or genetic inheritance,[2,5] or enteric injury associated with fetal varicella.[5] Yet, both the origin and the pathogenesis of CA remain unknown.

Conclusion

We report a case of colonic atresia in an infant who presented with typical signs of colonic atresia, absence of meconium passage and abdominal distention, and clinically suspected Rubinstein-Taybi Syndrome, which, to our knowledge, has not been reported in association with colonic atresia. The diagnosis of colonic atresia was swiftly confirmed by examination with barium enema.

Author Contributions

Conceptualization, T.R.H.; Acquisition, analysis, and interpretation of data, A.N.Y. and C.T.W.; Writing – original draft preparation, A.N.Y.; Review and editing, A.N.Y. and C.T.W.; Supervision, T.R.H. All authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. All authors had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis.

Disclosures

None to report.

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