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Anorectal malformation and Hirschsprung disease in an otherwise healthy infant

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ABSTRACT

Background: Hirschsprung disease and anorectal malformations are two conditions that are rarely associated with one another. Current reports of anorectal malformations with concurrent Hirschsprung disease predominantly describe children with genetic syndromes or other chromosomal anomalies. This report describes a case of imperforate anus and Hirschsprung disease in an otherwise healthy patient highlighting the importance of considering Hirschsprung disease in any patient with stooling difficulties following anorectoplasty for anorectal malformation.

1. Introduction

Hirschsprung disease (HD) and anorectal malformations (ARM) are two common congenital malformations infrequently associated with each other. There are fewer than 130 such cases reported in the literature with 2.4% of HD patients having ARM and 0.17% of ARM patients having HD [1]. Case reports of children with HD and ARM predominantly describe children with concomitant genetic syndromes or chromosomal anomalies [1–3]. ARMs are typically diagnosed at birth by physical exam and subsequent HD symptoms may be delayed by the diversion of stool and delay in definitive diagnosis. We present a case of a primary anoplasty performed shortly after birth with ongoing stooling issues and subsequent diagnosis of HD.

2. Case report

A Caucasian female was born via uncomplicated, spontaneous vaginal delivery at 39 weeks and 2 days. APGARS were 9 and 9 at 1 and 5 minutes, respectively. The patient presented to the author's institution at 2 days of life with abdominal distention, non-bilious vomiting, and failure to pass meconium. Physical exam revealed imperforate anus with perineal fistula at the anterior margin of the sphincter. An orogastric tube was placed with immediate return of large volume air and gastric fluid. Abdominal x-ray was obtained revealing diffuse gaseous distention of the small bowel with a small amount of gas in the rectum (Fig. 1). On day 3 of life, the patient underwent posterior sagittal anorectoplasty with electromyography of the anal sphincter. Postoperatively, the patient was able to pass stool spontaneously and formula feeds were gradually increased to goal. She was discharged on postoperative day 7.

At three weeks of age, the patient returned to the emergency department for inability to tolerate oral feeds with non-bilious emesis and infrequent, hard stools. Abdominal x-ray revealed marked gaseous dilation of bowel in the mid abdomen with concern for obstruction (Fig. 2). Physical exam revealed a distended abdomen and patent anus with no stenosis of the prior anorectoplasty. Contrast enema at one month of life showed decreased caliber of the rectum (Fig. 3) and subsequent suction rectal biopsy revealed

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aganglionosis. She was diagnosed with HD to the level of proximal sigmoid colon and underwent diverting colostomy at 7 weeks of age. At 5 months of age, the patient underwent laparoscopic-assisted Soave pull-through. Since that time, she has been stooling spontaneously and growing well.

3. Discussion

Hirschsprung disease (HD) and anorectal malformations (ARM) are two common congenital malformations seen in pediatric surgery. However, the two malformations are infrequently reported as coexisting anomalies [3]. The incidence of ARM ranges from 1:2000 to 1:5000 births, while HD is found in 1:5000 births [3–5].

The diagnosis of ARM is typically made shortly after birth with the absence of an anus in its normal position [4,6]. Types of ARM include low, intermediate, and high. Patients with ARM may present with or without fistula [3,4,6]. There are known associated conditions that occur with ARM such as VACTERL[A] and CHARGE[B].

HD is typically diagnosed in neonates with symptoms of abdominal distension and/or failure to pass meconium within 48 hours [3,5]. The gold standard for diagnosis of HD is a suction rectal biopsy revealing aganglionosis [7].

While ARM and HD are, separately, common anomalies seen in neonates, the two malformations are rarely reported together. According to a 2013 systemic review by Hofmann and Puri in *Pediatric Surgery International*, only a total of 90 cases of associated ARM and HD had been reported from 1952 to 2013 [3]. The systemic review revealed a high incidence of associated conditions in the cases when ARM and HD are coexistent: 11 cases with Currarino syndrome, 8 cases with Down syndrome, 3 cases with Cat eye syndrome, and 1 case with Pallister-Hall syndrome [3]. The patient described in this case is unique in that she has no other comorbidities.

In the case of coexistence of HD and ARM, there is often a delay in diagnosis of HD with a median diagnosis at 8 months of life [3]. This is thought to be due to the assumption that the symptoms of HD are related to complications after reconstructive surgery for ARM [3]. In this case, the diagnosis of HD was made at 1 month of age with rectal biopsy after presenting with inability to tolerate feeds and infrequent stools.

4. Conclusions

In conclusion, HD should be considered in all patients with continued constipation after reconstructive correction of ARM to avoid delay in diagnosis of HD.

Patient consent

Consent to publish the case report was not obtained. This report does not contain any personal information that could lead to the identification of the patient.

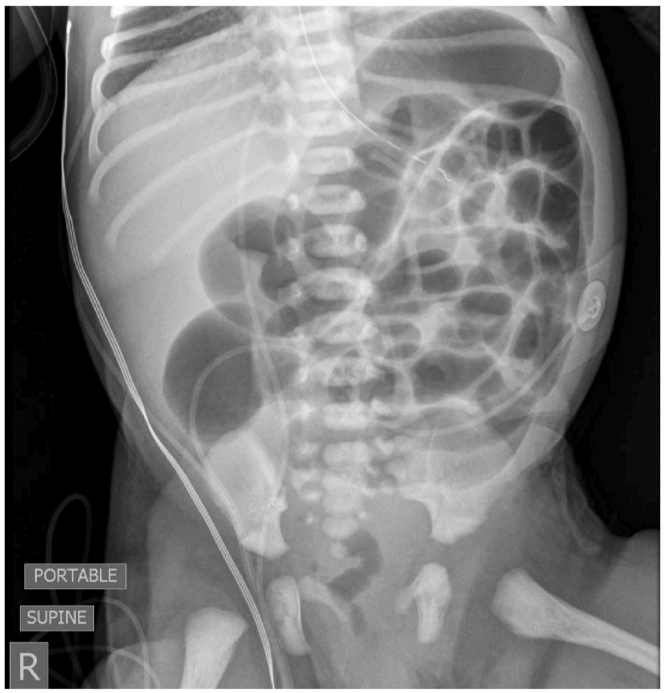

Fig. 1. Abdominal x-ray, day of life 2.

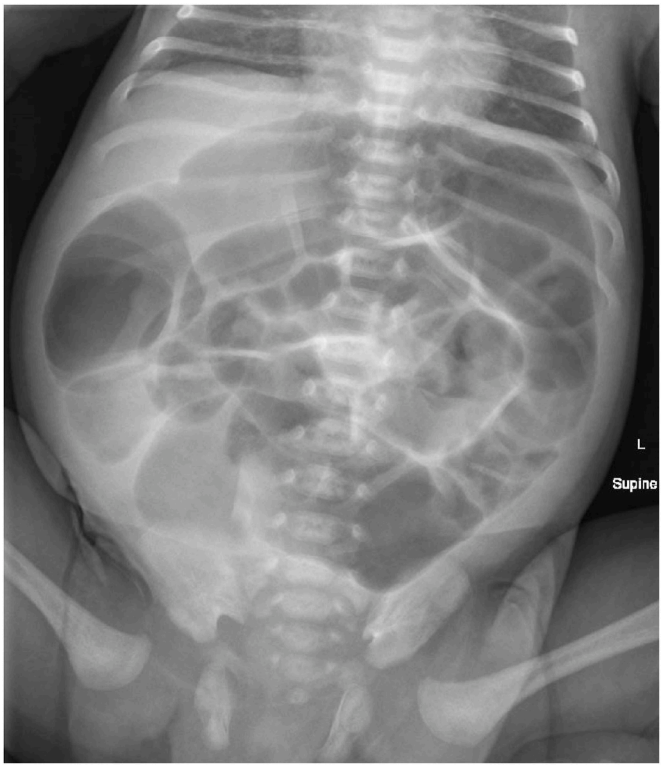

Fig. 2. Abdominal x-ray at one month of life.

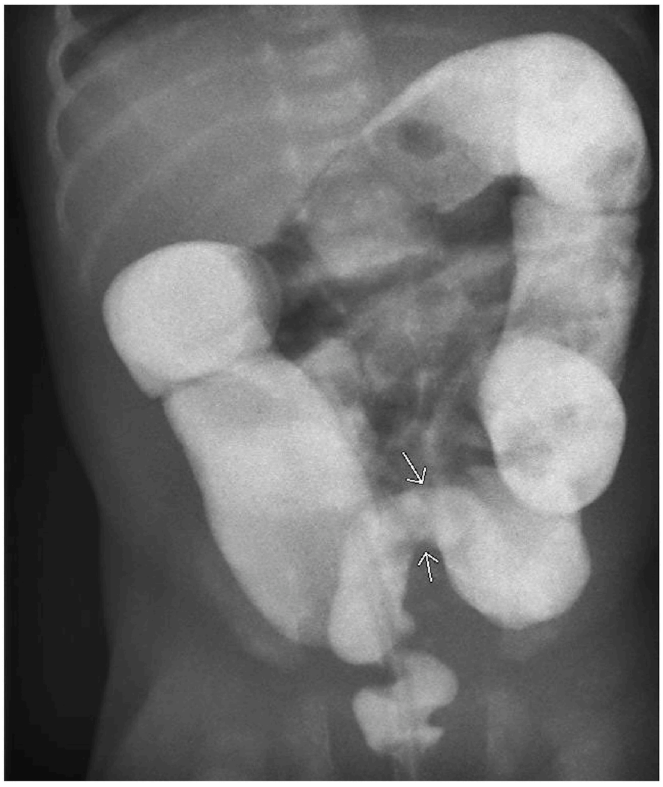

Fig. 3. Contrast Enema, one month of life.

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Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

Appendix

A VACTERL: Vertebral defects, Anal atresia, Cardiac defects, TracheoEsophageal fistula or esophageal atresia, Renal anomalies and Limb defects

B CHARGE: Coloboma of the eye, Heart defects, Atresia of the nasal choanae, Retardation of growth and/or development, Genital and/or urinary abnormalities, and Ear abnormalities or deafness

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