Isolated Extended long segment Hirschsprungs disease - A Diagnostic Dilemma?

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Abstract

Background: Isolated Extended long segment aganglionosis is one of the rarest forms of Hirschsprung's disease (HD). Establishing a correct diagnosis has proven to be challenging, because while the clinical and radiological features can be useful, but they are not pathognomonic.

Case report: We are reporting one more case to the existing literature, a girl baby who was admitted in S Nijalingappa medical college and HSK hospital with recurrent non bilious vomiting, abdominal distention, recurrent constipation. Baby did not respond to conservative measures. Explorative laporatomy was done, with working differential diagnosis of hirschsprungs disease, congenital bands, Small bowel stricture. On table findings were suggestive of extended long segment hirschsprungs disease. Diagnostic rate has progressively decreased as age advances in infants and the diagnostic rate being almost 40% by the age of 3 months. Extended long segment HD is challenging to diagnose radiologically.

Conclusion: Pre operative diagnosis is essential for isolated extended long segment HD, although often regarded as a relatively easy diagnosis, may be one of the most difficult diagnoses in Paediatric Surgery, mainly due to variations and difficulties in interpreting the relevant clinical, histological, and radiological findings.

Key words: Extended long segment Hirschsprung's disease; pre operative diagnosis; seromuscular biopsies; rectal biopsy.

Introduction

Isolated Extended long segment aganglionosis is one of the rarest forms of Hirschsprung's disease and until recent years it has been considered deadly[1]. Establishing a correct diagnosis has proven to be challenging, because while the clinical and radiological features can be useful, but they are not pathognomonic[2].

Case report

We report a case of a girl who was admitted in S. Nijalingappa medical college and HSK hospital with recurrent non bilious vomiting, abdominal distention, recurrent constipation. With an initial suspicion of ?congenital bands, ? Small bowel stricture, baby underwent explorative laporotomy with ileostomy and appendectomy. Diagnosis of Extended long segment hirschsprungs disease was established.

A 3months old female baby admitted with makedly distended abdomen, recurrent non bilious vomiting, recurrent constipation since 5days. There was past history of admissions for similar complaints which improved with conservative treatment at local hospital. Her routine blood investigations and thyroid profile within normal limits. Erect abdomen x-ray and ultrasound abdomen showed only dilated bowel loops, barium enema showed normal caliber colon and was inconclusive[fig 1]. Baby did not respond to conservative measures like nil peroral, Laxatives, enemas.

Then planned for explorative laporatomy, with working differential diagnosis of ?hirschsprungs disease, ?congenital bands, ? Small bowel stricture. On table findings (Fig 2) were suggestive of extended long segment hirschsprungs disease (large bowel was collapsed along with 15-20cms ileum and revealed massively dilated jejunum and ileal loops with normal bowel patency). Multiple seromuscular biopsies were taken from the recto sigmoid, proximal colon, distal ileum, ileotomy site, appendix and a loop ileostomy

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was done. Appendectomy was also performed. Post operatively, baby started doing well, abdominal distention and vomiting reduced, ileostomy started functioning on day 3 and baby was discharged on day 5. Rectal biopsy is the most confirmatory diagnosis for hirschsprungs disease. The histopathological examination revealed no ganglion cells in rectosigmoid, proximal colon, distal ileum and appendix. Ileostomy site shows ganglion cells in the myenteric plexus and no hypertrophic nerve bundles[fig 3]. Based on these findings, the diagnosis of extended long segment aganglionosis was established.

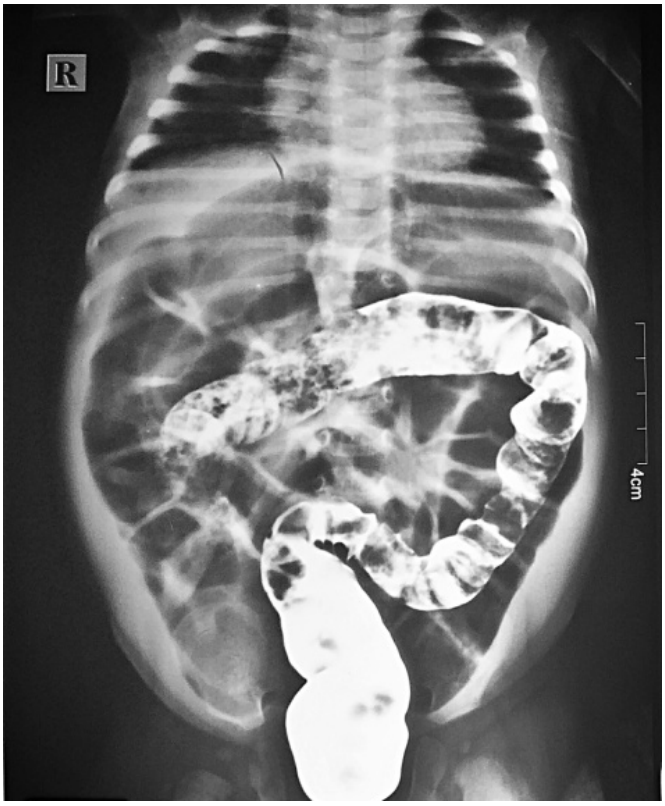

Figure 1: Barium enema – normal caliber colon which was inconclusive

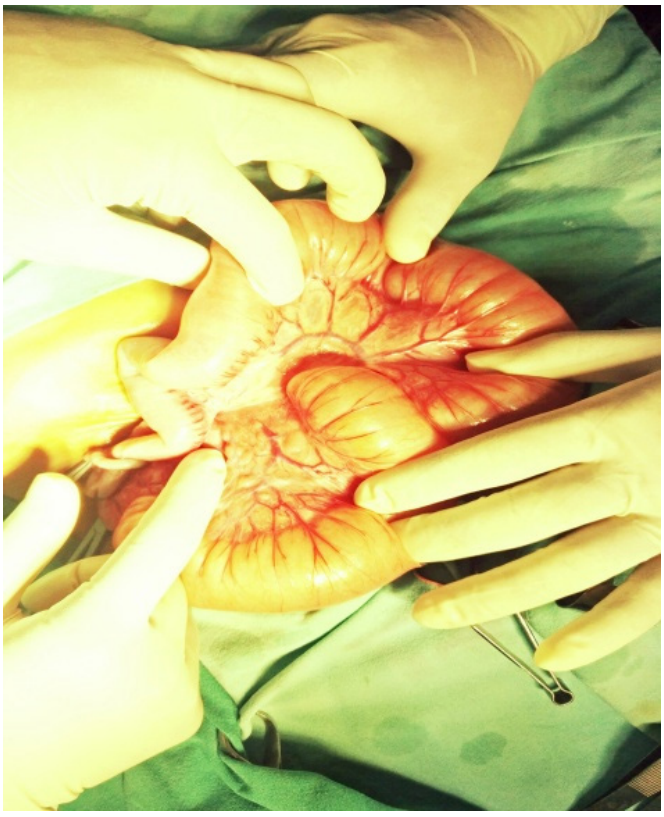

Figure 2: Intraoperative large bowel shows transitional zone.

Figure 3: Ileostomy site shows ganglion cells in the myenteric plexus and no hypertrophic nerve bundles

Discussion

Hirschsprung disease (HD), also known as 'congenital megacolon' is characterized by the absence of ganglion cells in the myenteric and submucosal plexuses of the intestine. Long-segment HD is usually defined as a transition zone which is proximal to the mid-transverse

colon. Total colonic aganglionosis (TCA)[3,4], involves aganglionosis involves the entire large intestine. Extended long segment hirschprungs disease (ELSHD): Defined as total colonic aganglionosis (TCA) with extended small-bowel involvement of ileum. Extended long segment aganglionosis represent the most extreme and rare form of congenital Hirschsprung's disease[5-7], being reported in only 3-12% of all HD cases. We are reporting one more case to the excisting literature. Diagnostic rate has progressively decreases as age advances in infants and the diagnostic rate being almost 40%[5] by the age of 3months. The newborn with HD is usually a full-term baby and presents with a distended abdomen, bilious vomiting and delay in the passage of meconium[6].

Our patient was born at 38 weeks gestation and presented abdominal distension, non bilious vomiting. Baby at neonatal period improved with symptomatic treatment. The diagnosis of HD is supported by family history with a higher incidence of the disease, clinically (early symptoms) and laboratory investigations, barium enema and it is confirmed by the histopathological examination[5]. The diagnosis of extended long segment aganglionosis is difficult compared to other forms of HD and often causes delays in initiating the appropriate treatment[5]. Rectal biopsy is the most confirmatory diagnosis for hirschsprungs disease.

Although multiple procedures exist for the treatment of extended long segment HD[8,9], there is no current consensus on a superior operative procedure. The treatment of extended long segment involves early decompression of the colon by an ostomy, using the most distal ganglionated bowel. This is usually followed by a second reconstructive procedure, after the child has had adequate time to grow and nutritional abnormalities have been corrected[9]. Early surgical procedures for the treatment of extended long segment aganglionosis have evolved from the procedures used for other forms of HD, such as the pullthrough procedures described by Swenson, Duhamel and Soave[8].

A nationwide survey of TCA and EA was performed between 1988 and 1992 in Japan. This survey reported a frequency of TCA of 4.5% (50 cases) and a frequency of EA of 5.2% (57 cases) among 1,121 cases of aganglionosis, and a male-to-female ratio for all types of aganglionosis of 3.4:1, and for TCA and ELSHD of 1.5:1[10].

Extended long segment HD is challenging to diagnose radiologically Campo et al. reviewed the radiological findings in 13 patients with TCA and concluded that there were no specific pathognomonic findings on barium enema studies[11]. In our case also barium study was inconclusive.

In our case, the first surgical intervention was explorative laparotomy with ileostomy with multiple seromuscular biopsies. Histopathological examination revealed ganglionic cells at ileostomy site.

We have planned for second reconstructive procedure after 1yr .

Congenital anomalies and conditions commonly associated with Hirschsprung Disease are Down syndrome (trisomy 21), Waardenberg–Shah syndrome, Yemenite deaf-blind-hypopigmentation, Goldberg–Shprintzen syndrome, Smith–Lemli–Opitz syndrome, Multiple endocrine neoplasia 2, Congenital central hypoventilation syndrome (Ondine'scurse)[12].

The Waardenburg–Shah syndrome is an autosomal recessive disease where Hirschsprungs' disease and the Waardenburg syndrome are present together. It is rare but Waardenburg–Shah syndrome patients have a higher incidence of total colonic aganglionosis with or without small bowel involvement[12].

Conclusion: Pre operative diagnosis is essential for isolated extended long segment HD, although often regarded as a relatively easy diagnosis, may be "one of the most difficult to diagnose in Paediatric Surgery", mainly due to variations and difficulties in interpreting the relevant clinical, histological, and radiological findings. Extended long segment agangliosis is a rare form that was considered fatal in infancy. Improvements in supportive care, early and accurate diagnosis and appropriate treatment have led to an increased survival rate in these patients.

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