# A rare case of total intestinal aganglionosis

**Dinushan Kaluarachchi**[1], **Ananda Lamahewage**[2]

*Sri Lankan Journal of Child Health,* 2013: **42**(1): 50-51

(Key Words: Hirschsprung disease; total intestinal aganglionosis)

## Introduction

Hirschsprung disease is a form of functional intestinal obstruction characterized by congenital absence of ganglion cells in the myenteric and submucosal plexuses of the distal colon. The extent of bowel involvement in this disease is variable[1]. Long segment disease involving entire bowel is very rare.

## Case report

A 45 day old baby boy was transferred to the Lady Ridgeway Hospital for Children, Colombo, Sri Lanka (LRH) with a complaint of non functioning jejunostomy from a Children's Hospital in United Arab Emirates (UAE). Baby was born to Sri Lankan parents living in UAE by vaginal delivery at 34 weeks of gestation with a birth weight of 2.04 kg. Meconium was not passed during the first 24 hours and at 28 hours of age baby developed bilious vomiting and abdominal distension. Abdominal x-ray showed dilated small bowel loops (Figure 1).

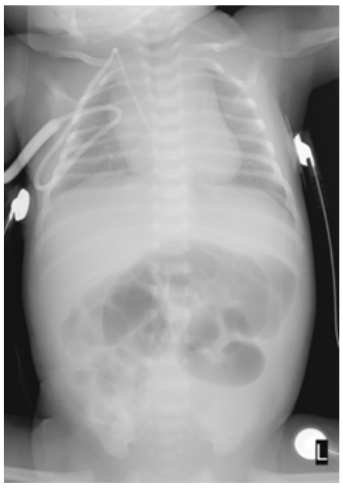

**Figure 1:** ***Abdominal x-ray***

[1]*Intern Medical Officer,* [2]*Consultant Pediatric Surgeon, Lady Ridgeway Hospital for Children Colombo, Sri Lanka*

(Received on 29 April 2012: Accepted after revision on 25 May 2012)

Upper gastrointestinal (UGI) contrast study was carried out and was suggestive of small bowel atresia (Figure 2)

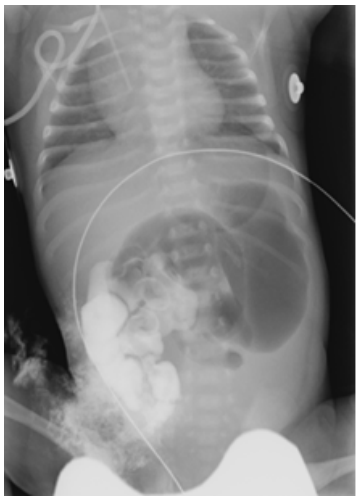

**Figure 2:** ***Upper GI contrast study***

Exploratory laparotomy was performed on day 2 of life but revealed no small bowel atresia. Proximal jejunostomy was created with the proximal stoma in right iliac fossa and the distal jejunostomy was placed as a mucus fistula at the lateral end of the laparotomy wound. Biopsies were taken from several sites (53, 93, 155 cm distal to duodeno-jejunal flexure, right colon and mid sigmoid colon). All the biopsies were negative for ganglion cells.

Due to persistence of symptoms and non-functioning jejunostomy, baby underwent second exploratory laparotomy on day 26 of life. This time 3 biopsies were taken and only the biopsy taken from the stomach showed ganglion cells. Histology was highly suggestive of total intestinal aganglionosis.

Child was started on parenteral nutrition. On day 45 of life baby was transferred to LRH for further management as requested by the parents. At LRH, UGI contrast study was performed and the duodenum was found to be markedly dilated; jejunal loops were also dilated but to a lesser degree than the duodenum. Contrast medium was seen to come out of jejunostomy confirming that it was patent.

Oral feeds were started once patency was demonstrated by the contrast study, but the feeds were tolerated poorly. Previous histology slides were brought down from UAE and reviewed by the consultant pathologist at LRH. Stomach biopsy showed normal ganglion cells but the rest of the biopsies failed to show ganglion cells. Ileostomy site biopsy was taken and it again was free of ganglion cells. Interestingly none of the biopsies showed hypertrophic nerves. At age of 68 days patient deteriorated and died due to sepsis. Autopsy was not performed due to parental concerns.

**Discussion**

Total intestinal aganglionosis accounts for less than 5% of patients with Hirschsprung disease[2]. Although a male preponderance is seen in most series of Hirschsprung disease, with a greater extent of aganglionosis, the sex ratio is closer to 1:1[3].

Presenting symptoms are that of upper intestinal obstruction. The only reliable means of diagnosis rests on serial biopsies once the condition is suspected. Previous reports[1] also have described a similar histological picture with absence of both ganglion cells and hypertrophic nerve fibers as in our case.

This condition has been considered to be universally fatal in series reported earlier[3]. New therapeutic options have extended the survival periods[4]. Therapeutic options for the management of total intestinal aganglionosis are long term parenteral nutrition, extended myectomy-myotomy[5] and intestinal transplantation[6].

**References**


1. Bodian M, Carter CO, Ward BCH. Hirschsprung Disease. *Lancet* 1951; **1**: 302-9. http://dx.doi.org/10.1016/S0140-6736(51)92290-8

2. Stringel G. Extensive intestinal aganglionosis including the ileum: a new surgical technique. *Journal of Pediatric Surgery* 1986; **21**: 667-70. http://dx.doi.org/10.1016/S0022-3468(86)80382-7

3. Canciano DA et al. Total intestinal aganglionosis. *Journal of Pediatric Surgery* 1985; **20**: 456-7. http://dx.doi.org/10.1016/S0022-3468(85)80241-4

4. Saxton ML, Ein SH, Hoehner J, Kim PC. Near-total intestinal aganglionosis: long-term follow-up of a morbid condition. *Journal of Pediatric Surgery* 2000; **35**: 669-72. http://dx.doi.org/10.1053/jpsu.2000.5939

5. Ziegler MM, Royal RE, Brandt J, et al. Extended myectomy-myotomy. A therapeutic alternative for total intestinal aganglionosis. *Annals of Surgery* 1993; **218**: 504-9. http://dx.doi.org/10.1097/00000658-199310000-00010

6. Sauvat F et al. Intestinal transplantation for total intestinal aganglionosis: a series of twelve consecutive children. *Journal of Pediatric Surgery* 2008; **43**: 1833-8. http://dx.doi.org/10.1016/j.jpedsurg.2008.03.028