

Paediatrician's Suspicion and Radiologist's Precision in Diagnosing Hirschprung's Disease in a Child: Case Report

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ABSTRACT

Hirschprung's disease (HD) is also called as aganglionic megacolon. The entity falls in the group of congenital motor disorders. The usual presentation is in the form of not passing of meconium, abdominal distension with or without vomiting. We present a 2-years old child who was brought with complaints of constipation and difficulty in passing stool. The child was evaluated radiologically by plain radiography, ultrasonography (USG) and barium enema and was diagnosed as a case of short segment type of Hirschsprung disease. The parents had been counselled and advised for pull-through surgery for the child. It is a dilemma for the paediatrician to confirm the diagnosis of Hirschprung's disease until a radiologically confirmed diagnosis is made. Many cases are delayed for the treatment because of non-diagnosis or delayed diagnosis. The importance lies in the early diagnosis for further surgical management.

Keywords: *Aganglionic Megacolon; Barium Enema; Congenital; Meconium; Pediatricians*

INTRODUCTION

Hirschprung's disease is also called as aganglionic megacolon of congenital origin and 20% falls in the familial category. This has got dominance pattern of inheritance which is of incomplete penetrance type. The incidence reported is 1:5000-8000 and prevalent in males. The short segment is the most common and the incidence increases with age. This is rarely seen in premature infants. The incidence is > 95% in full-term babies.¹ The common complaints are delayed meconium passing, loss of appetite, abdominal distension, vomiting and enterocolitis. There are a vast variety of children who are asymptomatic and

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15% of cases remain undiagnosed till 5 years of age. Some are diagnosed at adolescent and puberty. The early diagnosis is very important to avoid complications like enterocolitis and toxic megacolon. Danish paediatrician Harald Hirschsprung first described this entity in 1886 in the autopsy.²

CASE REPORT

2-years old boy was brought to the children outpatient department with complaints of chronic constipation, pain abdomen and loss of appetite. There was a history of the distended abdomen and passing stool once or twice a week (Figure1).



Figure 1: Photo of a 2-years old child with Hirschsprung's disease shows a distended abdomen

The complaints were of over one and a half years duration. The child was irritable, low energy activity as compared to other children of his age. There was no developmental delay. On examination, the child was of averagely built body constitution. The abdomen was mildly distended, soft and non-tender with visible prominent veins over it. The child was having microcytic, hypochromic anaemia. Systemic examination was unremarkable. The child was advised plain radiograph of the abdomen followed by ultrasonography. Plain x-ray abdomen had shown paucity of gas in the pelvis and colonic regions (Figure 2).



Figure 2: Plain x-ray abdomen shows scoliosis with concavity to the right side (blue

arrow). Small bowel loops show distension with the paucity of gaseous shadows in the pelvis region (red star)

USG examination was limited due to bowel gas. Because of keeping the clinical history and examination, there was a strong suspicion of Hirschsprung disease. The child underwent a barium enema study on unprepared bowel with restricted exposures to avoid excessive radiation exposure. There was evidence of a short aganglionic segment of the distal part of the large bowel (Figure 3a & 3b). Delayed film after 24hrs had shown still sufficient residual contrast within the dilated proximal colon (Figure 3c).



Figure 3: Barium enema study. **3a)** anteroposterior (AP) film shows the short narrow segment of the recto-sigmoid colon (yellow arrow). Dilated small bowel loops are seen in the rest of the abdomen (green star). **3b)** lateral radiograph shows the normal recto-sigmoid space. **3c)** Delayed AP barium enema film of abdomen. There is a lot of contrast left in the colon (red arrow) with the dilated colon (blue star) having a narrowed segment at the end (yellow arrow)

The diagnosis of short segment Hirschsprung's disease was made. The child underwent a rectal suction biopsy which was taken from above the dentate line which confirmed the diagnosis. The affected segment was aganglionic and there was hypertrophy of the nerve endings. The parents of the child had been counselled for the contemplation of surgery through the ileoanal pull-through operation.

DISCUSSION

Hirschsprung's disease comes in the category of congenital aganglionosis in the distal colon and rectum. Routinely, the majority of neonates pass meconium within 24 hours but

few having this pathology may not do because of the underlying defect. If meconium is not passed, there is suspicion of intestinal obstruction.³ Many conditions have to be kept in mind like Hirschprung's disease, meconium plug syndrome, small left colon syndrome, anorectal malformation, neuronal intestinal dysplasia, hypoganglionosis or some other congenital associated syndromes. This affects the cells in both myenteric and submucosal complexes. The exact theory is not understood whether this defect is because of non-migration or degeneration of neuroblasts. They present with a different type of clinical setting as per the type of the entity.⁴ The intrinsic nervous system of the gut is the enteric nervous system (ENS). In absence of ENS, there is no control of the propulsive motility of the gut. This leads to constriction as there is no neutrally mediated mechanism which leads to functional obstruction. This may lead to enterocolitis and perforation. Spontaneous perforation occurs in 3% of cases, especially in long segment variety.⁵ The final event is mortality because of all these factors. Diagnosis and management always remain challenging.⁶ Most of the neonates pass meconium in the first two days of life, but if not, then the other conditions including HD should be ruled out. The majority of cases present by five years of age and it is rare in adults. There are four different types of narrowed segments seen as per histopathological examination as given in Table 1.

Table 1. Different types of narrowed segments in Hirschprung's disease

TYPE	Region affected	% age of occurrence
Short segment	Rectum and sigmoid colon	75
Long segment	Extends up to the splenic and transverse colon	15
Total aganglionosis	Total	7.5
Ultra short segment	3-4 cm of internal anal sp	controversial

A plain abdominal x-ray can only show signs of intestinal obstruction in the form of air-fluid levels. A barium enema is done with great care in unprepared bowel. The transition zone has to be identified.⁷ This is a narrow segment with proximal dilatation. The narrowed segment shows fasciculation or saw-tooth appearance. The Rectosigmoid ratio is also important to take into cognizance which is always <1 in normal cases.⁸ Delayed films show the residual contrast within the dilated colon as it was seen in our case. The diagnosis can be picked up in antenatal ultrasonography examination. Low osmolality water-soluble contrast medium can be used for enemas where there is the fear of spillage of barium in the cases of perforations.⁹ Anal manometry and rectal biopsy are part of the hallmark of the investigation schedule. A full-thickness biopsy is always required for a definitive diagnosis. The new method for surgical management is resection of the narrowed segment with terminal-terminal colorectal anastomosis through the pull-through operation. The procedure was without any complications.¹⁰ Once the diagnosis is made precisely by the radiologist, the path becomes easier for pull-through surgery. The procedures can be modified as per the clear road map.

CONCLUSION

Hirschprung's disease required to be diagnosed early because of its known complications like enterocolitis, toxic megacolon and perforation. The disease remains undiagnosed in the beginning because of its subtle clinical symptomatology. The radiologist should diagnose at the earliest to avoid these complications. Surgical management and type depend upon the type of the disease and all require different types of surgeries. The diagnosis is multispeciality teamwork including histopathological confirmation.

CONFLICT OF INTEREST

None

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