Case Report

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# Hirschsprung's Disease in a Twin Neonate With One Suffering From Multiple Skip Segments

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**Citation:** Sangsari R, Kadivar M, Saeedi M, Mirnia K, Ghavami Adel M, Safavi M. Hirschsprung's Disease in a Twin Neonate With One Suffering From Multiple Skip Segments. Case Reports in Clinical Practice. 2021; 6(3):112-115.

**Running Title:** Hirschsprung's Disease in A Twin Neonate

## ABSTRACT

This is a new case, affecting a twin that both with Hirschsprung's disease, one was suffering from long-segment Hirschsprung's disease with skip segmented. Our surgeon suspected the absence of abnormal vessel tortuosity in the transitional zone; thus, the appendix was sent for permanent pathology. There were ganglion cells in the colostomy site but no ganglion was found in the appendix. Complete biopsies from different parts of the intestine demonstrated positive ganglion cells in the transverse colon, ascending colon, and the distal ileum, jejunum, and duodenum, and negative ganglion cells in the rectum, sigmoid, descending colon, appendix, and 5 cm far from the ileocecal valve were observed. Therefore, the Kimura procedure was done. The most important key in such cases is the surgeons' observation during operation.

**Article info:**
**Received:** 02 Jun 2021
**Revised:** 14 Jun 2021
**Accepted:** 24 Jun 2021

**Keywords:**
Hirschsprung's disease; Frozen sections; Meconium

## Introduction

The enteric neural system is a network that controls different gastrointestinal functions: blood flow, secretion, absorption, and motility. This complex system is derived from neural crests [1]. Human vagal Neural Crest Cells (NCC) emerge from the neural tube and migrate to the entire gut [2]. Vagal and sacral NCC should migrate to the gut and then survive, proliferate, differentiate, and finally form axons. This process leads to the formation of two major nerve centers: myenteric plexus (Auerbach's plexus) located between the external and longitudinal muscle layers and the

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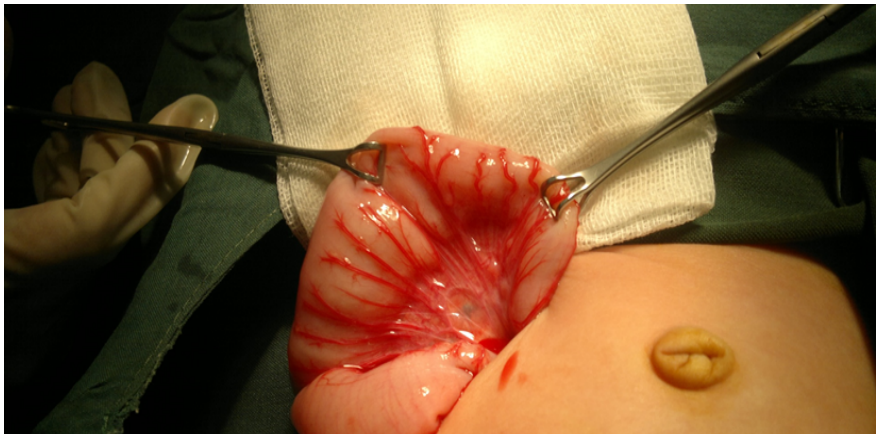

**Figure 1.** The appearance of the colon

The abnormal tortuous vessel was not seen at the transitional zone.

submucous plexus (Meissner's plexus). Many neurotransmitters are released from this plexus that control smooth muscle contraction and electrophysiological events [3]. This system is under the influence of many intrinsic factors, like genes and neuropeptides, and acquired factors, like inflammation, infectious, and immune-mediated etiologies; thus, any disruption in this pathway leads to aganglionosis and abnormal gut motility.

The main disorder of gut motility during neonatal life is Hirschsprung's disease that presents with no passage of meconium, abdominal distension, and bilious vomiting. Diagnosis is made by plain abdominal x-ray and Gastrografin enema and is confirmed by biopsies. In rare conditions, these procedures cannot show the precise level of the affected colon due to an abnormal scattering of ganglion cells during migration.

## Case Presentation

A 15-day-old male neonate with a gestational age of 38 weeks and weight of 2600 g resulting from twin delivery was admitted due to no passage of meconium and bilious vomiting. The baby was operated with a diagnosis of ileus meconium without any investigation in the first center. After the first surgery, clinical symptoms persisted; therefore, a second surgery was performed with a diagnosis of obstruction due to the adhesion band, but the clinical condition did not improve. The first-born twin (his brother) had similar milder symptoms without surgery in the first center. Thus, both neonates were referred to our hospital.

In contrast study, Hirschsprung's disease was confirmed in both babies. The first baby underwent surgery without colostomy and was discharged, and after 6 months of follow-up, he was fine. For the second baby, a contrast study showed a transition zone at the beginning of the descending colon. In laparotomy, there were no ganglion cells in the descending colon, and then, a colostomy was performed at the end of the transverse colon. Because the appearance of the colon was not compatible with Hirschsprung's disease, and the abnormal tortuous vessel was not seen at the transitional zone (Figure 1), an appendectomy was performed and sent for pathology. The results showed ganglion cells in the colostomy site, but no ganglion was found in the appendix. Besides, no improvement was observed in symptoms. Thus, due to the first diagnosis of ileus meconium, cystic fibrosis was considered and treatment

**Table 1.** Key messages

| What This Study Adds | What is Already Known |
|---|---|
| Abnormal vessel tortuosity at the transition zone in Hirschsprung's disease helps us to surgery.<br>Hirschsprung's disease can have a different manifestation in a twin delivery.<br>Hirschsprung's disease can involve the skip area of the intestine. | Surgeon observation of the intestine is less critical.<br>Hirschsprung's disease cannot be seen as a skip segmented in one twin.<br>Hirschsprung's disease involves the proximal of the intestine. |

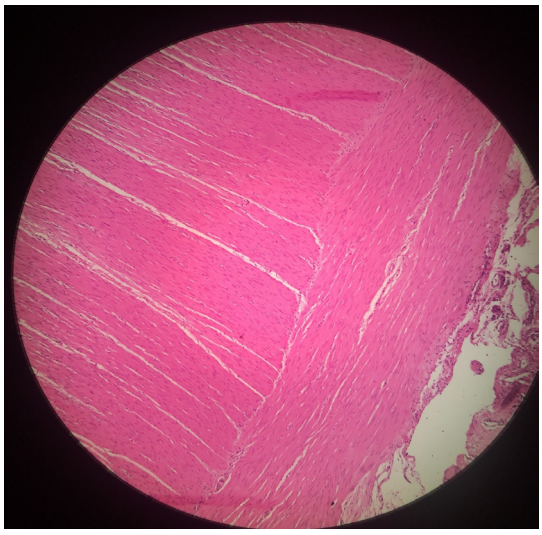

**Figure 2.** Specimen of the near the ileocecal valve

was performed, but abdominal distension and bilious vomiting continued and the genetic test for cystic fibrosis was negative.

In the upper GI follow-through study, the contrast reached to colostomy site after several days, and then, we prescript medical management for intestinal movement disorder, but it was not useful. Therefore, another laparotomy was performed with more extensive biopsies. The pathologists reevaluated the specimens for Neuronal Intestinal Dysplasia (NID), but it was negative. The result showed positive ganglion cells in the distal jejunum, ileum, and ascending and transverse colon, but it was negative for appendix, 5cm up the ileocecal valve, and descending colon till rectum (Figure 2).

To supply water and electrolytes, the Kimura procedure was performed and 20 cm of jejunum longitudinally to ascending colon was anastomosed and brought out in colostomy in the fifth surgery. Unfortunately, after this surgery, short bowel syndrome appeared, and with replacement fluid therapy and total parenteral nutrition, after three months, the baby's condition let us discharge him, but one month after discharge, the baby died due to enterocolitis.

## Discussion

This case was operated for five times, of which two cases were performed due to a medical error in the first center. He underwent three operations for his final treatment. Although it is hard to tolerate for a neonate, it is due to the complex nature of the disease, as it is a rare form of Hirschsprung's disease. We do not want to discuss genetics and pathology because obtaining the proper specimen and the surgeon's qualification can affect both. Besides, every hospital may not be able to ascertain genetic mutations. Our goal is to show when we suspect such rare cases. Hirschsprung's disease can be caused by genetic transmission [4] through Mendelian pattern, both dominant and recessive trait, with a male predominance [5]. Long-Segment Hirschsprung's Disease (LSHD) is seen in 21% of familial cases [6].

Our patient was an LSHD. In LSHD skip areas may be seen due to bypassing a segment of the colon by enteric cells [7, 8]. With a review of the history, our surgeon made an appendectomy during the first operation because she did not see abnormal vessel tortuosity at the zone. Nitin Pant et al. reported an aberrant 'corkscrew' vasa recta observed on the colonic surface in the area of the transition zone [9]. Lister for the first time reported these aberrant vessels [10]. The surgeon's observation during operation is important to perform more extensive biopsies from the colon till we do not miss rare pathologies (Table 1).

## Ethical Considerations

### Compliance with ethical guidelines

There were no ethical considerations to be considered in this research.


### Funding

This research did not receive any grant from funding agencies in the public, commercial, or non-profit sectors.


### Conflict of interest

The authors declared no conflict of interest.


### Aknowledgment

We would like to thank from NICU Nurses of Childrens Medical Center Hospital.


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