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Review Article

Contingency for Novel Diagnosis and Therapies for Hirschsprung's Disease

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ASTRACT

The majority of frequent reason of low functional blockage of the bowel in pediatrics is a Hirschsprung disease, which is brought on by a genetic mutation that disrupts the enteric nerve system. The absence of enteric ganglia over a varied length of intestine characterizes this developmental condition, which is classified as a neurocristopathy. Hirschsprung disease is a complicated surgical and medical condition that seems to have different social and health consequences based on the age of the patient and level of neurodevelopment. This review examines the genetic variables that influence development of Hirschsprung disease as well as its prevalence and congenital connection. An overview of a Hirschsprung's disease and its etiology, diagnosis, course of therapy are the goals of this article.

Keywords: Complication, Diagnosis, Etiology, Hirschsprung's disease, Management of disease and Ayurvedic treatment.

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INTRODUCTION

Hirschsprung disease is said to affect roughly 1 in 5000 live births globally. Hirschsprung disease instances are associated with a genetic abnormality in about 12% of cases. Boys are three times more probably than females to ancestry have Hirschsprung disease. Eighty percent of those who suffer from the illness do not have a Hirschsprung's disease. However, the child's likelihood of having Hirschprung disease is only 1% if one of the parents has it.

There is a 4% probability that a sibling born to a couple with Hirschsprung disease will also have the condition. There are less than a million instances annually in India (Figure 1). The loss of nerve cells in the colon and other large intestine muscles is a hallmark of Hirschsprung's illness, which makes it difficult to evacuate feces. Hirschsprung's disease is sometimes referred to as congenital megacolon. Most often, it is chronic constipation in babies. Hirschsprung's illness is identified if, within the first 24 to 48 hr of life, a newborn fails to pass meconium. Meconium is the baby's first stool after delivery.

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Internal Sphincter of the Anal where illness starts, and it spreads proximally over a variable amount of the intestines. The reason behind Hirschsprung's illness is several. Enteric Nerve System (ENS) is absent from the distal colon in this deadly birth abnormality that is partially penetrant, non-mendelian, and hereditary. This illness can manifest alone or in conjunction with other hereditary conditions like Waardenburg syndrome or Down syndrome. Hirschsprung's disease has a 2530% fatality rate.

Types

Short Segment Disease

In 80% of instances, aganglionosis only affects the recto-sigmoid colon.

Long Segment disease

In about 15% to 20% of patients, the aganglionosis spreads proximally to the sigmoid colon.

Total Colonic Aganglionosis

In about 5% of cases, Aganglionosis affects the entire large intestine.

Total Intestinal Aganglionosis

It is extremely uncommon.

Distinguishing between childhood functional constipation and Hirschsprung's Disease

Feature	Functional constipation	Hirschsprung's disease
Starting Point	two to three years	When born
Prolonged transit of meconium.	Infrequent	Ordinary
Fear of bowel movements.	Typical	Not often
Stool dimensions	Really big	Tiny, ribbon-shaped.
inadequate growth.	Rare	Common
Enterocolitis	Not ever	Feasible
Rectal ampulla	Expanded	Tapered
Barium enema	No transitional zone.	Transitional zone
Rectal biopsy	Regular	Absence of ganglion cells.

Etiology

- Aganglion cells in the colon and variable region of the rectum are absent from birth.
- It results from the interaction of two genes that encode different proteins.

1. RET proto-oncogene (chromosome 10).

2. EDNRB gene (chromosome 13).

Symptoms

Symptoms in newborn age

- Fail to pass meconium.
- Abdominal distension.
- Rectal tube cannot be put easily.
- Vomiting.
- Fever.
- Signs of dehydration.

Symptoms in infants

- Meteorism.
- Constipation.
- Ulceration, bleeding.
- Palpable fecaloma.
- Putrescent diarrhea.
- Anemia.
- Hyperproteinemia.

Symptoms in Childhood

- Dilated drum-like belly.
- Defecation in 7-10 days.
- Rectum is empty and narrow.
- Gracile limbs
- Multiple fecal masses.

Risk Factors

1. Children with Down syndrome.
2. It occurs 5 times more in males than in females.
3. Inadequate motility
4. Complex inheritance factors.
5. Having a sibling with Hirschsprung's disease.

History

Hirschsprung's Anniversary

About 150 years ago, Danish pediatrician Dr. Harald Hirschsprung (18301916) published the first explanation of Hirschsprung's disease. His writing of the first account of two children who perished from intestinal blockage known as "Congenital megacolon" is highly recognized. At Copenhagen, Denmark's Queen Louise Children Hospital, Dr. Hirschsprung was employed as a physician. A significant milestone happens in 2016 that could have a significant effect on pediatric surgery, pediatric pathology, and clinical pediatrics.

When the story started

Dr. Hirschsprungs began his medical profession with a special interest in children after receiving his degree in 1855. Between the years of 1880 and 1885; Hirschsprung approached two babies who had comparable clinical characteristics. The first child did not have spontaneous bowel motions and had intestinal issues that continued shortly after delivery.

Similar intestinal distension and a final diarrheal crisis were present in the second case. The continual therapy of information scarcity was implemented. Sadly, though, both kids passed away, and a postmortem was performed. The rectum was dilated during autopsy. However, there was some ulceration and a constriction of the intestinal loops. The term "Hirschsprung's disease" first widely used in 1916, just before the 20th century came to an end.

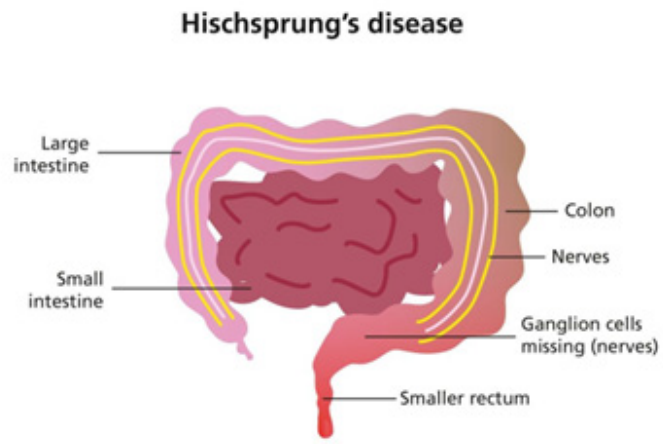

Figure 1: Hirschsprung's disease.

An Analysis of the Pre-Hirschsprung Era

According to a review of the scientific literature, between 1825 and 1888, there were about cases reported. Evidence suggests that the Hindu 20 Similar Surgeons of ancient India were aware of such illness. Similar to Hirschsprung's illness, Sushruta discusses a condition known as 98 Baddha nudo doram. An obstruction of the ano rectal canal is the cause of Baddha Gudodaram, according to Sushruta.

In Amsterdam, Netherlands, in 1891, Dutch anatomist Fredericus Ruysch reported a five-year-old girl who has stomach pain. It appears that the normative approach to treating stomach pain was ineffective. Practically speaking, there was no evacuation, and the infant died. Domenico Battini, an Italian physician from the 19th century, reported on a youngster who had chronic constipation for ten years. After the youngster passed away, an autopsy revealed that the colon had severely dilated. Dr. Battini may have succeeded in describing a megacolon instance over a century before Hirschsprung.

A seventeen-year-old child who had constipation from infancy was described by Ebers in 1836. Gee published the autopsy results of a 4-year-old child who had a sigmoid colon "spasm" in 1884. Dr. Hirschsprung passed away on April 11, 1916, but his contributions to pediatric surgery and pathology known as "Hirschsprungs Kramkheit" are unparalleled. He has left a wonderful legacy for kids. He provides the kids with a setting that might be peaceful.

Beyond the Hirschsprung's discovery

One in every 5,000 live births has this illness. The primordial neural Crest cells migrate caudally. When there is congenital aberrant nerve innervation of the intestine, the chance that the neural crest cells won't make it to the distal colon is reduced when migration is delayed. One example of a clinical manifestation in babies is failure to pass meconium within 24 hr. 98% of newborns pass meconium in less than 24 hr. The toxic megacolon has bile-stained stool and fever. Diarrhea, vomiting, shock, and distension of the abdomen. Radiology reveals a small distal segment with a fixed transition zone-level funnel-shaped dilatation. When the rectum is enlarged, electromanometry reveals that the Recto-Anal Inhibitory Reflex (RAIR) is absent. The reflexive relaxation of the internal anal sphincter following rectal distension is known as RAIR. RAIR (+) denotes the norm. Hirschsprung disease is indicated by RAIR (-). The last diagnostic procedure, a rectal biopsy, shows no ganglion cells and elevated acetylcholinesterase activity. Down syndrome affects about 1 in 20 children with Hirschsprung's disease.

Pathophysiology

When Hirschsprung disease spreads from the anus to different lengths throughout the large intestine, ganglionic cells are absent in the colon's mesenteric and submucosal plexuses. The enteric nervous system is created by the neural Crest cells of the vagal segment migrating to the vagus nerve and subsequently entering the foregut mesenchyme in a cranial to caudal orientation.

The most frequent cause of this illness is the arrest neuroblast, which produced during the migration of cells of the neural crest during weeks 8 to 12 of fetal development. Sometimes normal cell migration happens, but the neuroblast cannot mature normally because of incorrect differentiation, ineffective apoptosis, or poor proliferation. Failure of peristalsis and bowel movements results from the aganglionic segment remaining in a tonic condition. Proximal bowel dilatation and abdominal distension are caused by a buildup of feces in the recto-sigmoid area.

Aganglionosis prevents the rectum's feces from causing the internal sphincter of the anal to relax. Deterioration of layer of mucosa and decreased blood flow can result from elevated intraluminal pressure. Hirschsprung's disease complications and bacterial growth may result from this. This may result in sepsis and mortality if not detected in a timely manner.

EXAMINATION

The diagnosis of Hirschsprung disease will be made after a thorough physical examination and series of tests.

Contrast Enema

The contrast enema test makes use of contrast-added enema solution and X-ray images. This imaging study the enema is quite beneficial in determining whether a child has Hirschsprung disease. The colon's characteristics are more visible on an X-ray when the solution is applied.

Abdominal X-ray

A colon blockage may be visible on a stomach X-ray. This test is the first step in diagnosing Hirschsprung disease. This disease cannot be accurately diagnosed with it.

Biopsy of the Rectal Area

Rectal biopsy testing is used to get the final diagnosis. In newborns, the care team performs a "Suction" rectal biopsy at the hospital room bedside. This procedure entails removing a sample of rectum cells for a pathologist to examine under a microscope. At the biopsy site, sensory nerves are absent. Thus, there is no pain associated with the surgery. A child's absence of ganglion cells is used by the pathologist to confirm that the child has Hirschsprung disease. A general anesthetic is used during a surgical biopsy.[34]

Anal Manometry

Anal pressure is measured by this test. The hospital bedside is where it can be done. Moreover, it examines the rectum's typical reflexes.

The anus has two tiny muscles that regulate bowel motions. The two muscles in question are the exterior and internal sphincters. To stop bowel movement leakage that is not under control, these muscles are contracted. For bowel movements to occur, these muscles need to open and relax simultaneously. One test to look at how these muscles work is anal manometry.

TREATMENT

Surgeons use surgical techniques such pull-through, ostomy, and Soave procedures to treat Hirschsprung disease.

Pull through procedure

Using a pull-through approach, a surgeon excises the section of the large intestine that lacks nerve cells. The surgeon then attaches the healthy section of the big intestine to the anus. Pull-through treatments can be performed laparoscopically or openly by surgeons. To do laparoscopic surgery, surgeons make small incisions in the abdomen and then insert a laparoscope and additional tools.

During open surgery, surgeons make a larger incision to reveal the abdomen. Sometimes a pull-through procedure is performed using equipment inserted through the anus rather than cutting the abdomen.[27]

Ostomy surgery

During ostomy surgery, doctors construct a stoma on a child's belly and connect it to the large and small intestines. After ostomy surgery, waste or excrement will leave the body through the stoma instead of the anus. The stoma is only transient. Later, surgeons can close the stoma, connect the anus to the healthy section of the intestine, and allow stool to pass. Ostomy surgery performed for megacolon, perforation, or severe Hirschsprung-associated enterocolitis.[27]

Sodium supplementation

Long segment Hirschsprung disease patients can require a supplement of salt. A straightforward urine test is done to determine a child's salt levels.

Biofeedback Therapy

The pelvic floor muscles, which regulate bladder and bowel motions, are strengthened and more coordinated with the aid of biofeedback therapy.

Soave Procedure

The soave technique was initially used in 1960. A pull-through colon is inserted inside the aganglionic muscle's cuff after the mucosa and submucosa of the rectum are removed during the Soave operation.

Duhamel Procedure

The Duhamel procedure was introduced in 1956. The normal colon is introduced retrorectally through the bloodless plane that divides the sacrum from the rectum during this procedure. When the aganglionic colon is cut into the rectum, the normal proximal colon and rectum are brought together. Two walls combine to generate a new lumen that is innervated posteriorly and ganglionic anteriorly. This technique offers the benefit of minimal pelvic dissection and a large anastomosis, which reduces the risk of stricture.[24]

Ayurvedic Management

Ayurvedic medicine can be used to treat the patient in this scenario that has Hirschsprung disease. There is a lower risk of side effects while using the Ayurvedic approach to treat any condition. Ayurvedic diagnostic Pakvasayagata vata was mentioned in vatavyadhi (musculoskeletal illnesses). Abdominal pain, Antrakoojana (bowel sound), issues with stools and urine passages, Pakvasayagata vata manifests as Trikvedana and Anaaha (abdominal distension).

Treatment for udavarta, or abdominal distension brought on by constipation and other factors, is identical to that for Pakvasayagata vata. Varti (suppository), Svedana (sudation). Hirschsprung illness is treated with abhyanga [massage] using shita jwarokta oil, snehana, virechana (light purgation), and the use of Anulomak meals as a carminative. Matra basti, Maidu Swedana (gentle sudation), and Mridu abhyanga (gentle massage) are also used to treat patients.

Drakshaveleha and Vrihatavatachintamani ras are medications prescribed for Hirschsprung's illness; Dabur, an Indian company, makes these. Daksha Jaiphala, Ela, Javitri, Vanslochan, Lavanga Tejapatta, Dalchini, and Kamalgatta make up Drakshaveleha. Gandhak, Raupya, Abhraka, Swama, Moti, Lauha, and Parad make up Vrihatavatachintamani.[8,9]

CONCLUSION

About 1 in 5000 live births suffers from Hirschsprung disease. It's a genetic illness. In boys, this ailment is three times more prevalent. Hirschsprung disease has a 1% risk of passing down to a kid if one parent has it.

It is challenging to pass feces because Hirschsprung's disease causes the colon to lose nerve cells. The internal anal sphincter is where this sickness begins, and it progresses proximally throughout the gastrointestinal tract. Numerous methods exist for diagnosing diseases, including contrast enema, biopsy, abdominal X-ray, and anal manometry. Hirschsprung disease cannot be accurately diagnosed by abdominal X-ray. Surgeons use surgical techniques such ostomy, pull-through, and save procedures to treat this illness.

Patients with Hirschsprung disease suffer fewer injuries after laparoscopic aided pull through. Rectal suction biopsy is an inexpensive, quick, and safe treatment. Both simplicity and accuracy are very good.

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CONFLICT OF INTEREST

The authors declare that there is no conflict of interest.

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