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Rare case of hirschprung disease

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Abstract

Introduction: Hirschsprung disease is a developmental disorder characterized by absence of ganglia in the distal colon, resulting in a functional obstruction.

Case Report: Here we are presenting a case report of *Hirschsprung Disease* in a viable age of fetus and sharing our experience in diagnosis and treatment of this rare entity.

Conclusion: Hirschsprung disease is diagnosed in the newborn period, but this rare entity is detectable at early stage with routine ultrasound investigations.

Keywords: Fetus, GIT, Aganglionic mega colon, Congenital anomaly, Hirschprung disease, Neuroblast

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Introduction

Hirschsprung disease is a developmental disorder characterized by absence of ganglia in the distal colon, resulting in a functional obstruction.

Most cases of Hirschsprung disease are diagnosed in the newborn period. Hirschsprung disease should be considered in any newborn that fails to pass meconium within 24-48 hours of birth. Although contrast enema is useful in establishing the diagnosis, full-thickness rectal biopsy remains the criterion standard. Once the diagnosis is confirmed, the definitive treatment is to remove aganglionic bowel and to restore continuity of the healthy bowel with the distal rectum, with or without an initial intestinal diversion.

Case Report

A 20 year old unbooked primi gravida presented with c/o of labour pains at 28 weeks of gestation with no h/o leaking. On examination her vitals are stable other findings were within normal limits. Systemic examination reveals no abnormality in respiratory, cardiovascular and central nervous system. The per abdomen examination showed 28 weeks relaxed gravid uterus with singleton fetus in cephalic presentation in non-engaged position with fetal heart rate of 144/min. Per vaginal examination reveals 3 cm cervical dilatation and well effezed cervix, with vertex presentation, and adequate pelvis. Her investigations were within normal limits.

With routine ultrasound anomaly scan fetal movements were present and fetal heart activity was visualized. Placenta was anterior and liquor was adequate. Foetus was presented with *microcephaly*, *vermian hypoplasia*, *hyperechogenic bowel* and *large bowel obstruction.* Other organs and foetal biometry was normal. (Fig. 1)

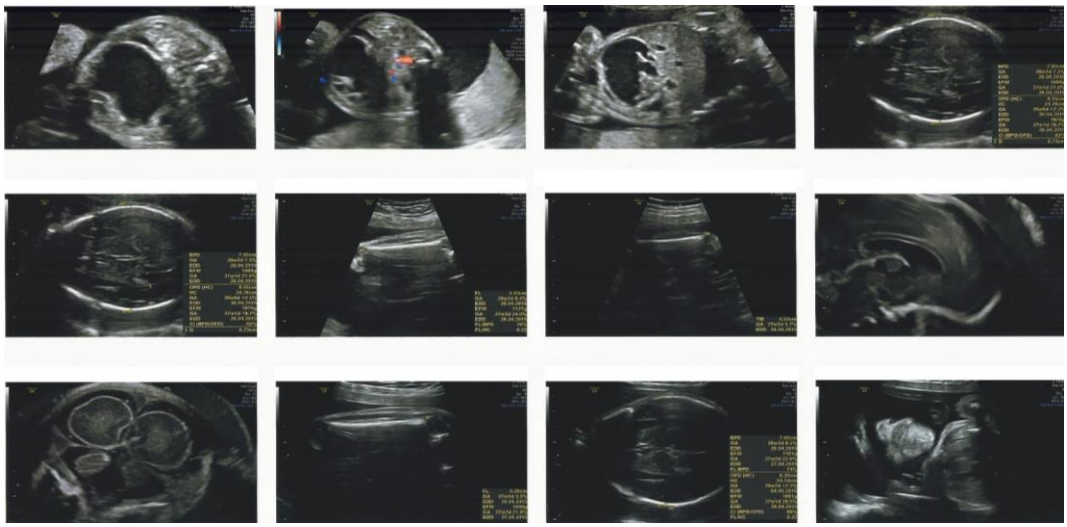


Fig. 1

Patient delivered a preterm baby weighing 900 gms. Cord blood was taken and sent for chromosomal study. Neonatal death. Baby send for Autopsy.

Foetal Autopsy study revealed severe adhesions in abdomen, distended 3rd part of duodenum and early part of jejunum. Rest of the findings was normal. (Fig. 2)

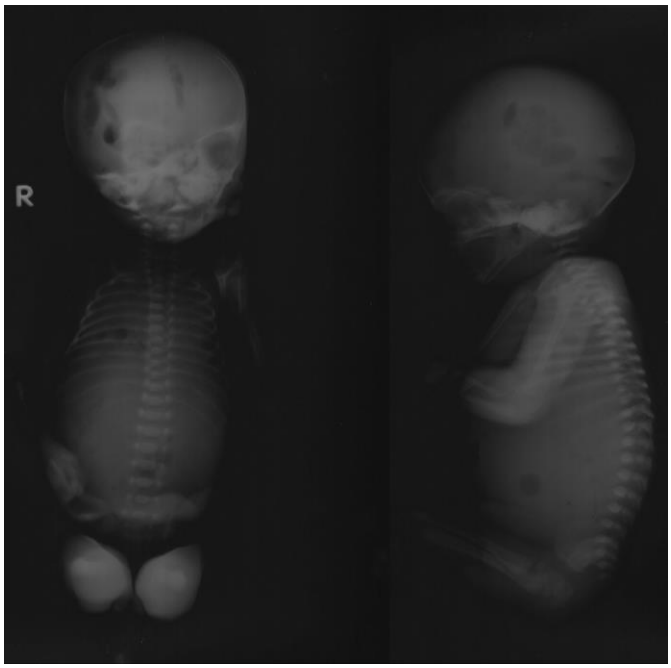
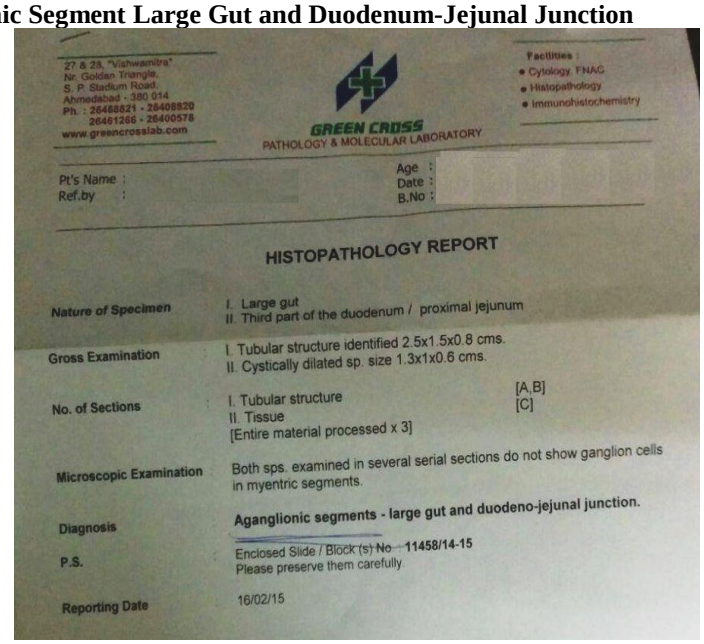


Fig. 2

On histopatho examination:

Report shows **Aganglionic Segment Large Gut and Duodenum-Jejunal Junction**



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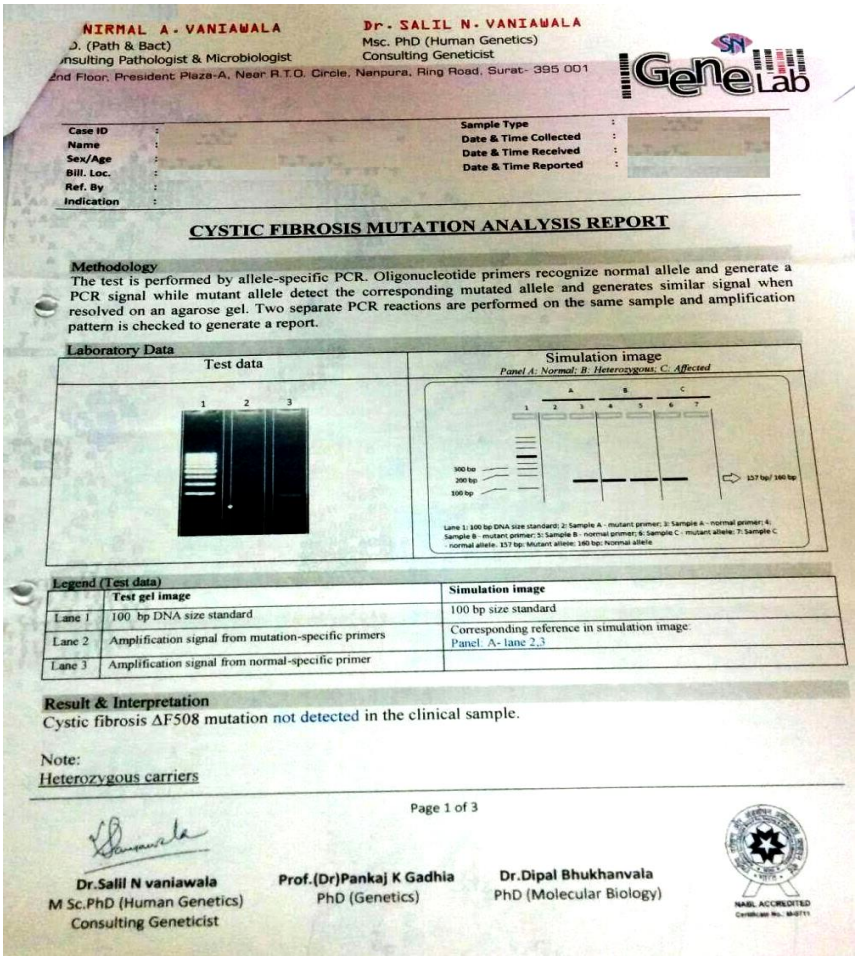
HISTOPATHOLOGY REPORT

Nature of Specimen	I. Large gut II. Third part of the duodenum / proximal jejunum
Gross Examination	I. Tubular structure identified 2.5x1.5x0.8 cms. II. Cystically dilated sp. size 1.3x1x0.6 cms.
No. of Sections	I. Tubular structure [A,B] II. Tissue [C] [Entire material processed x 3]
Microscopic Examination	Both sps. examined in several serial sections do not show ganglion cells in myentric segments.
Diagnosis	**Aganglionic segments - large gut and duodeno-jejunal junction.**
P.S.	Enclosed Slide / Block (s) No - **11458/14-15** Please preserve them carefully.
Reporting Date	16/02/15



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Case ID :
Name :
Sex/Age :
Bill. Loc. :
Ref. By :
Indication :

Sample Type :
Date & Time Collected :
Date & Time Received :
Date & Time Reported :

CYSTIC FIBROSIS MUTATION ANALYSIS REPORT

Methodology
The test is performed by allele-specific PCR. Oligonucleotide primers recognize normal allele and generate a PCR signal while mutant allele detect the corresponding mutated allele and generates similar signal when resolved on an agarose gel. Two separate PCR reactions are performed on the same sample and amplification pattern is checked to generate a report.

Laboratory Data

Test data

Simulation image
Panel A: Normal; B: Heterozygous; C: Affected

Legend (Test data)

	Test gel image	Simulation image
Lane 1	100 bp DNA size standard	100 bp size standard
Lane 2	Amplification signal from mutation-specific primers	Corresponding reference in simulation image: Panel. A- lane 2,3
Lane 3	Amplification signal from normal-specific primer	

Result & Interpretation
Cystic fibrosis ΔF508 mutation not detected in the clinical sample.

Note:
Heterozygous carriers

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Chromosomal Study shows *No Abnormality* in chromosomes 13, 18, 21 and sex chromosomes.

Discussion

Hirschsprung disease also called *congenital megacolon* or *congenital aganglionic megacolon*, is a form of megacolon that occurs when part or all of the large intestine or antecedent parts of the gastrointestinal tract have no ganglion cells and therefore cannot function. In Hirschsprung's disease, the migration is not complete and part of the colon lacks these nerve bodies that regulate the activity of the colon. The affected segment of the colon cannot relax and pass stool through the colon, creating an obstruction.[1] The stomach and esophagus may be affected, too.

Most cases of Hirschsprung disease are diagnosed in the newborn period. Hirschsprung disease should be considered in any newborn that fails to pass meconium within 24-48 hours of birth. Although contrast enema is useful in establishing the diagnosis, full-thickness rectal biopsy remains the criterion standard. Once the diagnosis is confirmed, the definitive treatment is to remove aganglionic bowel and to restore continuity of the healthy bowel with the distal rectum, with or without an initial intestinal diversion. This condition occurs in 1 out of every 5,000 live births.[2]

This condition occurs more frequently in boys than in girls, with as many as four boys affected for every girl. About 10% of cases are familial Also, children with Down syndrome have a substantially higher risk of having Hirschsprung disease.

Some cases of Hirschsprung disease can be related to a genetic (inherited) cause. Hirschsprung disease is associated with chromosomal abnormalities or syndromes in approximately 9% of cases.[10]

There is an increased chance that a couple will have a child with Hirschsprung disease if one of the parents has the condition, and the chance is higher if it is the mother who has the condition.

If a family has a child with Hirschsprung disease, there is a 3 percent to 12 percent chance that another baby will also have it.

Enteric ganglion cells are derived from the neural crest during embryonic development. In normal development, neuroblasts are found in the esophagus by the fifth week of gestation, and they migrate to the small intestine by the seventh week and to the colon by the twelfth week.[3] One possible etiology of Hirschsprung disease is the arrest of aboral neuroblast migration. Alternatively, although normal cell

migration may occur, neuroblasts may be subject to apoptosis, failure of proliferation, or improper differentiation within the affected distal intestinal segment. Fibronectin, laminin, neural cell adhesion molecule (NCAM), and neurotrophic factors present in the intestinal stroma are necessary for normal enteric ganglion development, whereas their absence or dysfunction may also have a role in the etiology of Hirschsprung disease.[4,5,6]

Hirschsprung disease is associated with chromosomal abnormalities or syndromes in approximately 9% of cases.[7]

The most accepted theory of the cause of Hirschsprung is that there is a defect in the craniocaudal migration of neuroblasts originating from the neural crest that occurs during the first 12 weeks of gestation. Defects in the differentiation of neuroblasts into ganglion cells and accelerated ganglion cell destruction within the intestine may also contribute to the disorder.[4]

This lack of ganglion cells in the myenteric and submucosal plexus is well-documented in Hirschsprung's disease.[3] With Hirschsprung's disease, the segment lacking neurons (aganglionic) becomes constricted, causing the normal, proximal section of bowel to become distended with feces. This narrowing of the distal colon and the failure of relaxation in the aganglionic segment are thought to be caused by the lack of neurons containing nitric oxide synthase.[3]

Prenatal ultrasound demonstrating bowel obstruction is rare, except in cases of total colonic involvement.[8]

Children with only a short segment of intestine that lacks normal nerve cells may not show symptoms for several months or even years. While individuals experience a range of symptoms, but most common are- Not having a bowel movement in the first 48 hours of life, Gradual marked swelling of the abdomen, Gradual onset of vomiting, Fever, Children who do not have early symptoms may present with Sepsis (overwhelming infection), Constipation that worsens over time Small, watery stool, Loss of appetite, Delayed growth.

Treatment of Hirschsprung's disease consists of surgical removal (resection) of the abnormal section of the colon, followed by reanastomosis. The first stage of treatment used to be a reversible colostomy. Enterocolitis, chronic obstruction, incontinence, constipation, and late mortality may occur late after surgery. Rectovesical fistulas have also been reported in the literature.[9]

Possible complications of surgery include anastomotic leak (5%), anastomotic stricture (5-10%), intestinal obstruction (5%), pelvic abscess (5%), and wound infection (10%). Long-term complications mostly affect patients with long-segment disease. They include chronic obstructive symptoms, incontinence, chronic constipation, enterocolitis, and late mortality. Although many patients encounter one or more of these problems postoperatively, long-term follow-up studies have shown that greater than 90% of children experience significant Improvement.[11] Patients with a syndromic association and those with long-segment disease have poorer outcomes.[12,13,14]

Conclusion

The incidence is 1 out of every 5,000 live births. Though it is a very rare entity it is detectable at early stage with routine ultrasound investigations. With this case we want to encourage the surgeons and physicians for early clinic-radiological diagnosis. We also want to for chromosomal study by cordocentesis, not all but in patients, where one of the parents has the condition, and the chance is higher if it is the mother who has the condition.

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