

A Rare Case of Extensive Intestinal Atresia Involving Small and Large Bowel

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Abstract: Hereditary multiple intestinal atresia (HMIA) is the rarest form of intestinal atresia, first reported by Winter and Zeltzerin 1956. In this case report we are presenting a pre-term male baby born at 35 weeks of gestation. The patient did not pass meconium. He had dark green vomiting and epigastric fullness. There was rectal atresia at about 3 cm from the anal verge. On laparotomy there was atresia at and beyond the Duodenojejunal junction, the distal bowel being extremely small in caliber and the whole intestine was absolutely solid with no lumen, involving both the small and large intestine right up to the anal canal. The bowel was non-canalized and solid with no mucosa at all. Enterotomy was done at different levels to check the patency of the gut but everywhere there was no lumen and even the large bowel including the rectum was atretic. Biopsy report revealed multiple microscopic lumens with all the microscopic layers. Immunological work up revealed severe immunodeficiency.

Keywords: Hereditary multiple intestinal atresia, severe combined immunodeficiency, Rectal atresia, calcification, duodenal obstruction, neonatal intestinal obstruction

1. Introduction

Hereditary multiple intestinal atresia (HMIA) is the rarest form of intestinal atresia, very scantily reported in the medical literature. It was first reported by Winter and Zeltzerin 1956.^{1,2} Later on an autosomal recessive pattern of inheritance was described in 1973 by Guttman et al.³ In 2014 mutations of the tetratricopeptide repeat domain-7A (TTC7A) was proposed by Fernandez et al.⁴ In HMIA the atresia involves an extensive part of the intestine which may occur at multiple locations throughout the small and large intestines. No part of the intestine is exempt to HMIA and it may involve any part of gastrointestinal tract. So far this condition is lethal even in those patient in whom resection is performed. The presence of severe combined immunodeficiency is considered as an essential association of HMIA^{1,5}. The condition commonly presents at birth with intestinal obstruction.⁶ The cause of death in these patients is twofold, one is total parenteral nutrition (TPN) dependence and secondly immunodeficiency making the patient vulnerable to acquire infection and septicemia ultimately leading to death. The proposed ultimate treatment is bowel transplantation which is still at experimental stage.

2. Case Report

A pre-term male baby born at 35 weeks of gestation, through spontaneous vaginal delivery (SVD) to a primigravida healthy 19-year-old mother. Father is 24 years old. Pregnancy was a product of consanguineous marriage. Antenatal work up showed massive polyhydramnios and double bubble sign suggestive of duodenal obstruction. No family history of any neonatal

intestinal obstruction. Post-natally the patient had a high gastric aspirate of dark green color and did not pass meconium. On abdominal examination there was fullness in the epigastrium but rest of the abdomen was soft and lax, no palpable mass and no visceromegaly. On active aspiration of the nasogastric tube there was a lot of air and green color sticky gastric aspirate due to which the epigastric fullness disappeared. On rectal examination the anal canal was seen patent but there was rectal atresia as the tube was not going beyond 3 cm into the rectum. Plain x-ray abdomen showed dilated stomach with paucity of gases in the distal bowel. There was a line of calcification on the right side of the abdomen (Figure 1).



Figure 1: (Double bubble sign with a thin line of calcification in the right lower quadrant)

A provisional diagnosis of complicated meconium ileus/duodenal obstruction was established. After

stabilization the baby was operated. At laparotomy there was atresia at and beyond the Duodenojejunal junction. The distal bowel was extremely small in caliber. The whole intestine was absolutely solid with no lumen at all. This was noticed right from the Duodenojejunal junction up to the rectum. The whole small and large bowel was non-canalized and solid with virtually no evidence of mucosal tissue at all. An attempt was made to open the gut (enterotomy) at different levels to check the patency of the lumen but everywhere the situation was the same. Even the large bowel including the rectum was found atretic. Not even a single centimeter of intestine had any lumen or mucosa.

Only some thick cheesy material which was recovered on opening the inside of the small intestine, coinciding with the calcification as seen on plain x-ray. Biopsy of mid-ileum segment was taken and sent for histopathology. Immunological work up revealed severe immunodeficiency. Chromosomal analysis was requested; however our facility lacks the resources. Histopathology report revealed multiple microscopic intestinal lumens. Each lumen has all the components of the wall of the intestine with intact tissue architecture, normal mucosal pattern and the muscle layer. The number of the intestinal lumens was four (4) in most of the microscopic fields Figure 2.

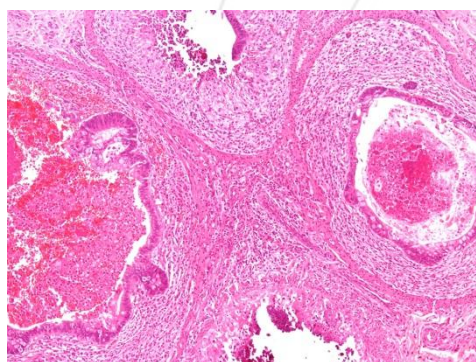


Figure 2: Histopathology (Note the four lumens each showing all the components of the wall)

The patient remained admitted for 3 months. He was receiving total parenteral nutrition through a central line but due to immunodeficiency he developed sepsis and died at the age of 3 months.

3. Discussion

Hereditary multiple intestinal atresia (HMIA) is the rarest form of intestinal atresia. It was first reported by Winter and Zeltzer in 1956.¹² It showed an autosomal recessive pattern of inheritance which is first described in 1973 by Guttman et al.³ The likely gene leading to this condition was proposed by Fernandez et al in 2014 to be deleterious mutations of the tetratricopeptide repeat domain-7A (TTC7A).⁴ Patients commonly present at birth with symptoms and signs of intestinal obstruction such as abdominal distension, epigastric mass and persistent bilious vomiting. Almost all those who were diagnosed with this condition died during early infancy especially in the first four months of life. Atresia can occur at multiple

locations throughout the small and large intestines and it may involve any part of gastrointestinal tract. However, this condition is lethal even in those patients in whom resection of the atretic part is performed.^{1,5} The cause of mortality besides short bowel is sepsis secondary to severe combined immunodeficiency and different other anomalies such as malrotation, cystic dilatation of bile duct, omphalocele, cardiac anomalies and congenital cystic adenomatoid malformation of lung.⁶ Karyotypic Abnormality Association in the living child with this condition is yet unreported.⁷ The proposed theories suggest that this condition may occur either at sites of obliterative embryonic events such as the stage of atrophy of vitelline duct or due to the failure of normal recanalisation of the solid cord stage of the fetal bowel leading to atresia of an extensive segment of the intestine, and lastly due to underlying fetal vascular accident.⁸ Familial intestinal atresia classified in conjugation with known embryological development into class 1 (pyloric atresia being a foregut anomaly), class 2 (duodenal atresia which occurs at the junction between the foregut and midgut), class 3 (HMIA affecting the entire gastrointestinal tract), class 4 (apple-peel atresia affecting the mid gut only) and class 5 (colonic atresia being a hindgut anomaly).⁹ As proposed by Boyd et al in 1994 this condition can be diagnosed antenatally during routine ultrasound scanning by evidence of bowel dilatation at 17 weeks of gestation and history of polyhydramnios which is considered as presenting feature in 20-35% of cases.¹⁰ There are no survivals reported so far in literature with HMIA but laparotomy is still considered necessary to confirm the diagnosis.⁶ The only hope for survival of patients with such condition is bowel transplantation which is still experimental and needs further research.

4. Conclusion

A high index of suspicion should be raised in those newborns who present with a triad of duodenal (or pyloric) atresia, rectal atresia and calcification on plain x-ray of abdomen. The only possible treatment of HMIA is bowel transplantation which is not available in our set up.

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