International Journal of Science and Research (IJSR)
ISSN (Online): 2319-7064
Index Copernicus Value (2015): 78.96 | Impact Factor (2015): 6.391

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 presentinga 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 atabout 3 cm from the anal verge. On laparotomythere 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 the 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.[12]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 gastointestinal 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 birthwith 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 ofgestation, through spontaneous vaginal delivery (SVD) to a primigravida healthy 19-year-oldmother. Father is 24 years old. Pregnancy was a product of consanguinous marriage. Antenatal work up showed massive polyhydromniosand 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).

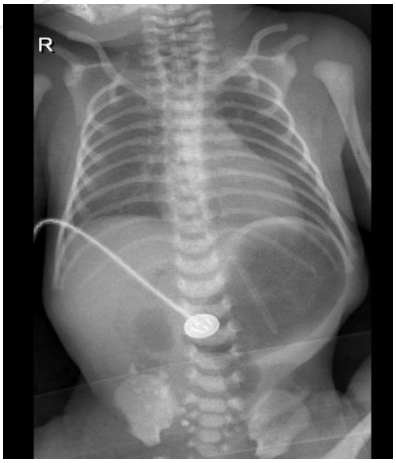

Figure 1: (Double bubble sign with a thin line of calcification in the right lower quadrant)

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

stabilization the baby was operated.At laparotomythere was atresia at and beyond the Duodenojejunaljunction. 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 foundatretic. 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-ilium 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 lumens 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 wasfour (4) in most of the microscopic fields Figure 2.

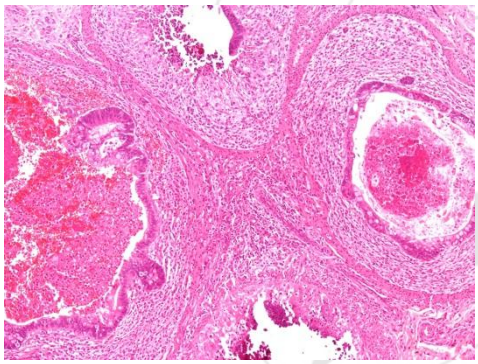

Figure 2: Histopathology **(**Note the four lumens each showing all the components of the wall**)**

The patient remained admitted for3 months. He wasreceiving 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 Zeltzerin 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 alin 2014 to bedeleterious 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 andpersistent bilious vomiting. Almost all those who were diagnosed with this condition died during early infancyespecially inthe first four months of life.Atresia canoccur at multiple locations throughout the small and large intestines and it may involve any part of gastointestinal tract.However, this condition is lethal even in those patient in whom resection of the atretic partis performed.[1,5]The cause of mortality besides short bowel is sepsis secondary to severe combined immunodeficiency and differentother anomalies such as malrotation, cystic dilatation of bile duct, omphalocele, cardiac anomalies and congenital cysticadenomatoid malformation of lung.[6]karyotypicAbnormality 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 tothe 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), class4 (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.

References

[1] Fernandez I, Patey N, Marchand V, et al. Multiple intestinal atresia with combined immune deficiency related to TTC7A defect is a multiorgan pathology: study of a French-Canadian-based cohort. Medicine (Baltimore). Dec 2014;93(29):e327.
[2] Winter ST, Zeltzer M. Congenital atresia of the ileum in two brothers; case report. J Pediatr. Aug 1956;49(2):194-196.
[3] Guttman FM, Braun P, Garance PH, et al. Multiple atresias and a new syndrome of hereditary multiple atresias involving the gastrointestinal tractfrom stomach to rectum. J Pediatr Surg. Oct 1973;8(5):633-640.
[4] Raj P SS, Ramji S, Sarin YK. Hereditary Multiple Gastrointestinal Atresia associated with Choledochal Cyst: A Rara Entity with Management Dilemma. Journal of Neonatal Surgery. 2014;3(3).

[5] Qureshi MF, Abdellatif M, Hindi MYA, Davis G, Qureshi S. Hereditary Multiple Gastrointestinal Atresia: A Report of three cases from the kingdom of Saudi Arabia. The internet journal of pediatrics and neonatology. 2013;16(1).
[6] Bilodeau A, Prasil P, Cloutier R, et al. Hereditary multiple intestinal atresia: thirty years later. Journal of Pediatric Surgery. 2004;39(5):726-730.
[7] Fatemeh Mahjoub, Kadivar M, Moradi B, Tabarzan N. Two Consecutive Stillbirths with Multiple Intestinal Atresias in a 30-year-old female. Iran J pediatr. 2009;19(4):435-438.
[8] Ali YA, Rahman S, Bhat V, Al Thani S, Ismail A, Bassiouny I. Hereditary multiple intestinal atresia (HMIA) with severe combined immunodeficiency (SCID): a case report of two siblings and review of the literature on MIA, HMIA and HMIA with immunodeficiency over the last 50 years. BMJ Case Rep. Feb 09 2011;2011.
[9] Shorter NA, Georges A, Perenyi A, Garrow E. A proposed classification system for familial intestinal atresia and its relevance to the understanding of the etiology of jejunoileal atresia. J Pediatr Surg. Nov 2006;41(11):1822-1825.
[10] Boyd PA, Chamberlain P, Gould S, Ives NK, Manning N, Tsang T. Hereditary multiple intestinal atresia--ultrasound findings and outcome of pregnancy in an affected case. Prenat Diagn. Jan 1994;14(1):61-64