Pyloric atresia with total small bowel atresia: a case report

Yosor Yaser Dabbour

Abstract: Intestinal atresia is a reason leads to intestines obstruction in the neonatal period . the multiple gastrointestinal atresia is a rare.We here report a case of pyloric atresia with total small intestinal atresia combined with immunodeficiency syndrome. We reported here about a case of pyloric atresia with total small intestinal atresia combined with immunodeficiency syndrome. The parents of the case are consanguineous and have other had one previous girl 4 years old which diagnosed with ITP and bronchia asthma. we have reviewed the clinical course and outcome in this article. The existing literature on multiple intestinal atresia, pyloric atresia with immunodeficiency is also reviewed.

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Introduction

An atresia is a congenital disorder of a hollow viscous which lead to complete hindering of the lumen (1). Intestinal atresia is a well-known reason of bowel obstruction in the newborn (2). The incidence rate of intestinal atresia is estimated with 1.3 to 3.5 per 10,000 live births, about 20% of intestinal atresia are associated with a chromosomal anomaly (1).

In recent decades, the management of neonates with intestinal atresia has been advanced because of improvements in intensive care of neonatal, total parenteral nutrition (TPN) using, operative technique, and anesthesia of neonatal (2).

Pyloric atresia (PA) is an uncommon malformation, it estimated to be responsible for less than 1% of gastrointestinal atresia, the incidence of pyloric atresia is 1 in 100,000 live births (3,4). Pyloric atresia presents as symptoms of gastric outlet obstruction. it's difficult to be characterized, but it may be accompanying with epidermolysis bullosa and other gastrointestinal anomalies like duplications (4).

Commonly, intestinal atresia occurs as an isolated lesion which has a good prognosis, but in this case, we represent a pyloric atresia with total small intestinal atresia combined with immunodeficiency syndrome which is incompatible with life.

Keywords: Atresia, Intestinal atresia, Bowel obstruction, Pyloric atresia, Newborn, Chromosomal anomaly, Epidermolysis bullosa,

Case presentation:

A 2 months old girl born by SVD at -+37 weeks, small for gestational age and low birth weight “1990 gm”. Admitted to a neonatal unit at

KASCH, Riyadh, KSA for low birth weight and to rule out intestinal atresia.

This girl had antenatal ultrasound findings which includes “dilated bowel loops and polyhydramnios”

There is consanguinity relation between her parent, her father is 33 years old and the mother is 24 years old. They had one previous girl 4 years old which diagnosed with ITP and bronchia asthma.

The mother not known to have any chronic disease, medications during pregnancy or history of infections. No psychosocial problems or related family history. Developed polyhydramnios at the third trimester and delivered her baby spontaneously vaginally at 37 weeks.

Physical examination of the infant was good: pink with no dysmorphic features, no abnormality detected, palate intact and nares patent. Apgar score was 9 on the first and fifth minutes.

- Chest examination was clinically free with bilateral equal air entry and no abnormality detected.

- Cardiovascular examination showed S1+S2+heart murmur.

- Abdominal examination showed mildly distended abdomen with no hepatosplenomegaly and patent anus.

- Neurological examination showed weak sucking, strong crying, normal tone and positive moro reflex.

- Musculoskeletal examination was clinically free.

The patient was kept NPO, gave antibiotics and IV fluids, and run some investigations. Routine investigations CBC and chemistry were within normal values for age. X-rays including KUB showed distended stomach with no air seen distal to that, as showed in figure 1.

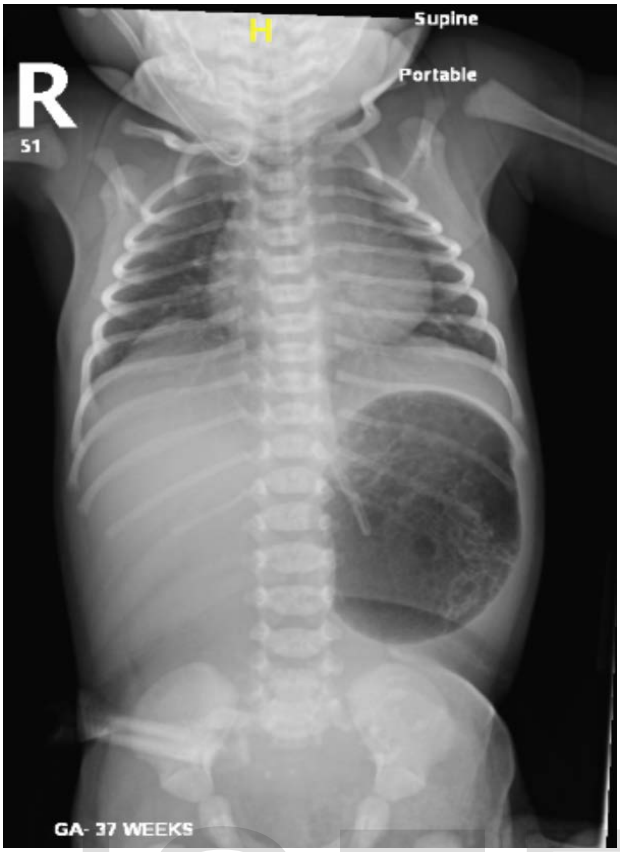

Figure 1: X-rays including KUB

Abdominal ultrasound showed fluid-filled dilatation of the duodenum and proximal small bowel loops. Also showed cystic fusiform dilatation of the CBD associated with dilatation of CHD and to lesser extend cystic duct.

Echo study documents PM-VSD, 2 small muscular VSD, large apical VSD, PDA, prominent left coronary.

The genetic diagnosis of gastrointestinal defects and immunodeficiency syndrome was confirmed.

Multiple other studies were done to rule out other anomalies and all were normal.

The patient was operated on 28-8-2017 and the finding was dilated duodenum, with total non-canalized small bowel up to the transverse colon with beaded appearance. All the bowel from duodeno-jejunum to transverse colon was atretic. There was septum in D1 and multiple septa in D4.

Pyloroplasty, jejunostomy and ileostomy was done before closure of the laparotomy.

The appendix, proximal small bowel, ileum and cecum were all submitted to pathological study and showed obliterated lumen with presence of ganglion cells confirming multiple intestinal atresia as showed in figure 2,3,4

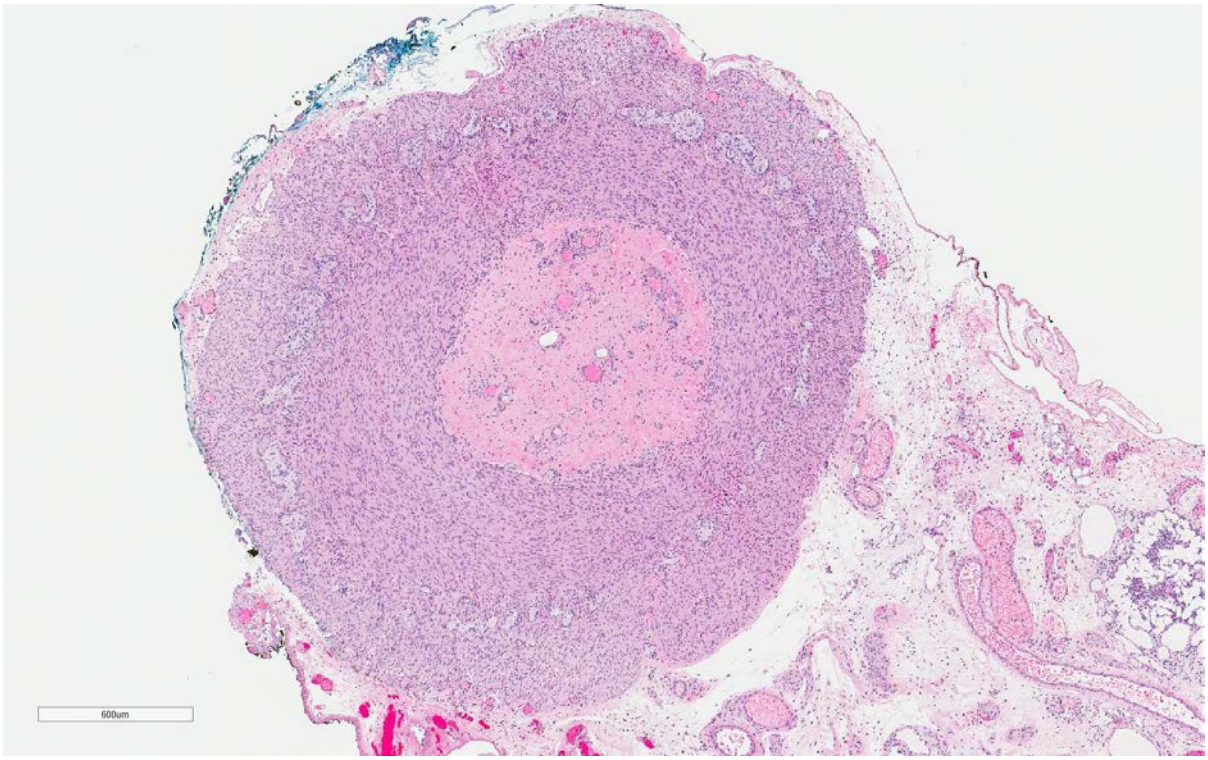

Figure 2: Cross section from intestine with lumen obliterations (atresia).

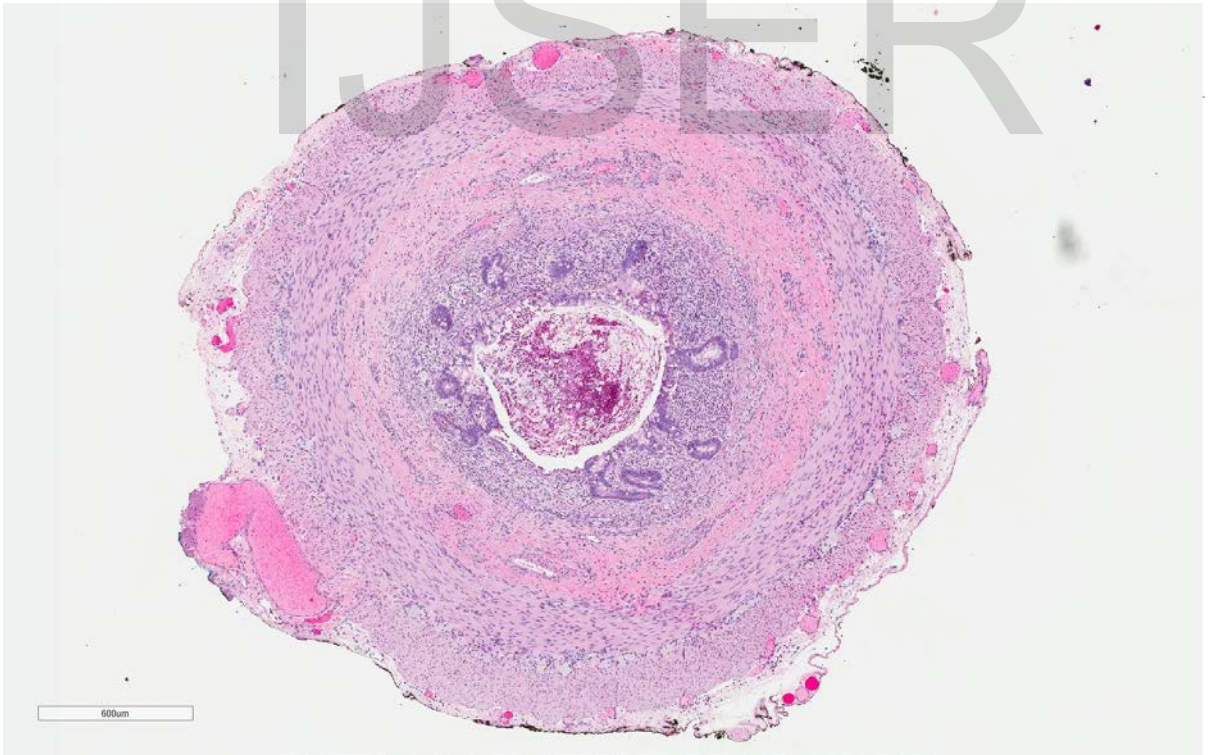

Figure 3: Other areas show lumen narrowing.

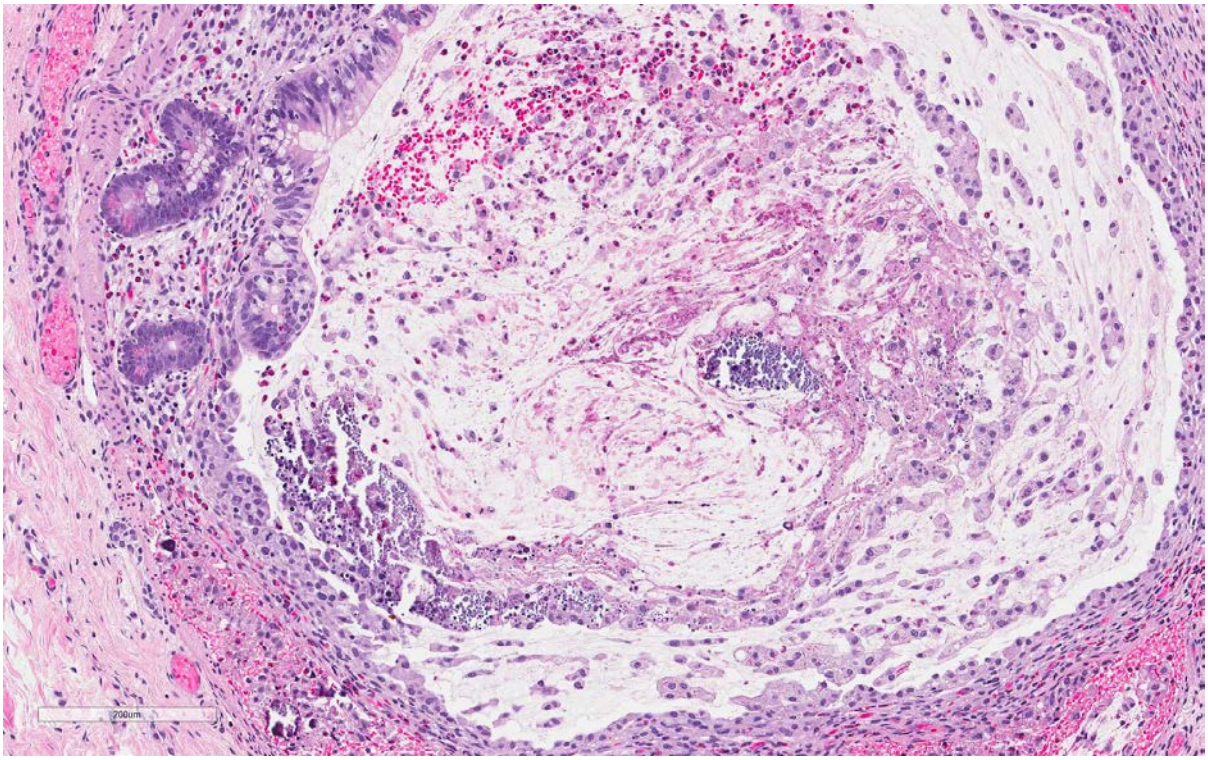

Figure 4: lamina propria eosinophilia, thick mucus in lumen and dystrophic calcification.

Discussion

Intestinal atresia represents about one-third of all neonatal intestinal obstruction cases. The reasons, diagnosis, clinical presentation, operative management, care after operations and outcome considerably differs depending on position of obstruction (5).

About 30%–55% of children with pyloric atresia are found to have other congenital anomalies–most commonly additional intestinal atresia

or epidermolysis bullosa (6). In this study we presented a case of a child with multiple intestinal atresia associated with immunodeficiency syndrome.

The exact etiology of atresia remains unknown (6). About 20% of intestinal atresia are associated with a chromosomal anomaly (1). The atresia could be due to a vascular insult or developmental arrest, others suppose that the fibrous membrane forms in response to chemical or mechanical irritation (6)

Pyloric atresia should be take into account in any newborn who demonstrate clinical manifestations of intestinal obstruction and non-bilious vomiting (6). The diagnosis in the present with pyloric atresia and multiple intestinal atresia case is confirmed with an abdominal radiograph which shows a single gastric bubble and a paucity of intestinal air as showed previously in figure 1. There is a delay in diagnosis and treatment repeatedly. Endoscopic and ultrasound exploration could be helpful (7).

In the present patient the genetic diagnosis of gastrointestinal defects and immunodeficiency syndrome was confirmed. Another study conducted in Saudi Arabia found three siblings suffering from Hereditary multiple Intestinal Atresia (HMIA) and Severe Combined Immunodeficiency Syndrome (SCID), The parents of these infants are first cousins (8). Hereditary multiple atresia (HMIA) including the gastrointestinal tract starting from pylorus to rectum are considered the most uncommon intestinal atresia form, the inheritance type was mentioned to be autosomal recessive. The inheritance of the severe combined immunodeficiency syndrome could be autosomal recessive or X-linked (9).

Our patient suffering from weak sucking, its well-known that intestinal atresia especially duodenal atresia lead to impair the swallowing. Also, an intestinal atresia in the fetus can lead to increased production of amniotic fluid (10). Our patient record showed that the mother developed polyhydramnios at the third trimester of her pregnancy

38% of the cases of Congenital anomalies of the abdominal wall or gastrointestinal tract are correlating with congenital heart disease (11). Other associated anomalies are cardiac in this patient, Echocardiogram suggests a Posterior Malalignment of Ventricular Septal Defect, 2 small muscular VSD, a large apical VCD and a Patent Ductus Arteriosis PDA.

For our patient pyloroplasty, jejunostomy and ileostomy was done before closure of the laparotomy. But, the presence of this child's immunodeficiency is a problem that makes her susceptible to intestinal inflammation. Ali et al. reported that The combination of HMIA and SCID is invariably lethal (5).

Present improvement in genetics and fetal medicine have made it possible to diagnose multiple intestinal atresia and immunodeficiency before birth. The termination decision consists on the local legal, social and cultural values in addition to the beliefs and wishes of parents. Families are less likely to opt for elective termination in Muslim countries including KSA, so in these countries the incidence

neonatal mortality due to lethal congenital malformations is high notably. In non-conservative societies, prenatal diagnosis and termination of fetuses with multiple intestinal atresia and immunodeficiency could be a preferable practical option (5).

Conclusion

Intestinal atresia is a known cause of intestinal obstruction in newborns, The surgical option is effective in the treatment of intestinal atresia but its efficiency is affected by other health problems that's associated with intestinal atresia such as immunodeficiency and cardiac anomalies. In the case of multiple intestinal atresia, attention should be paid to the possibility of associated immunological disorders and cardiac anomalies.

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