ECRONICON OPEN ACCESS

EC PAEDIATRICS
Case Report

Management of a Prenatally Diagnosed Congenital Anomaly and Comparison of Management in Developed and Developing Countries: A Case Report

Noela Olomi[1]*, Masatoshi Yamaguchi[1], Kazuhiko Nakame[2], Shinji Katsurag[1] and Fredy Rutachunzibwa[3]

[1]Department of Obstetrics and Gynecology, Faculty of Medicine, University of Miyazaki, Japan

[2]Department of Surgery, Division of Gastrointestinal, Endocrine and Pediatric Surgery, Faculty of Medicine, University of Miyazaki, Japan

[3]Department of Radiology in Kairuki University, Tanzania

***Corresponding Author:** Noela Olomi, Department of Obstetrics and Gynecology, Faculty of Medicine, University of Miyazaki, Kihara, Kiyotake, Miyazaki, Japan.

Received: August 02, 2024; **Published:** August 12, 2024

Abstract

Introduction: Congenital anomalies are structural or functional anomalies that occur during intrauterine life. These conditions may be identified before birth or after.

Case Report: A 23-year-old female was referred to University of Miyazaki Hospital. Ultrasound showed polyhydramnios, intrauterine growth restriction, single umbilical artery and dilated fetal bowel loops. Numerical chromosomal anomalies were ruled out by amniotic fluid analysis. The baby was born at 35 weeks, abdominal X-ray showed features of small intestinal atresia. On day two of life the baby underwent surgery and had a good prognosis.

Conclusion: Prenatal diagnosis is important to ensure timely management of anomalies.

Keywords: *Congenital Anomalies; Prenatal Diagnosis*

Background

Congenital anomalies as defined by the World Health Organization are structural or functional anomalies that occur during intrauterine life. They may be identified before birth, at birth, or later in life. Intestinal atresia is a congenital defect in which there is a complete blockage or lack of continuity of the bowel. Ileal and jejunal atresia are usually described collectively as jejunoileal atresia (JIA). JIA accounts for 1 in 5,000 to 1 in 14,000 live births, over one-third of affected children are born prematurely, there is no sex predominance, and usually there are no associated chromosomal abnormalities [1]. Despite the evolution of prenatal screening, use of ultrasound remains a vital and essential tool for prenatal screening of congenital anomalies [2].

Case Presentation

A 23-year-old female was referred to University of Miyazaki Hospital at 30 weeks gestation with fetal growth restriction, and suspected gastrointestinal atresia. Ultrasound showed fetal growth restriction, single umbilical artery (SUA), polyhydramnios, multiple bubble sign and a dilated stomach and bowel loops, suggesting gastrointestinal atresia (Figure 1). Amniocentesis was performed followed by chromosomal analysis. An arm-to-arm inversion of chromosome 5 was found. Numerical chromosomal abnormalities were not detected.

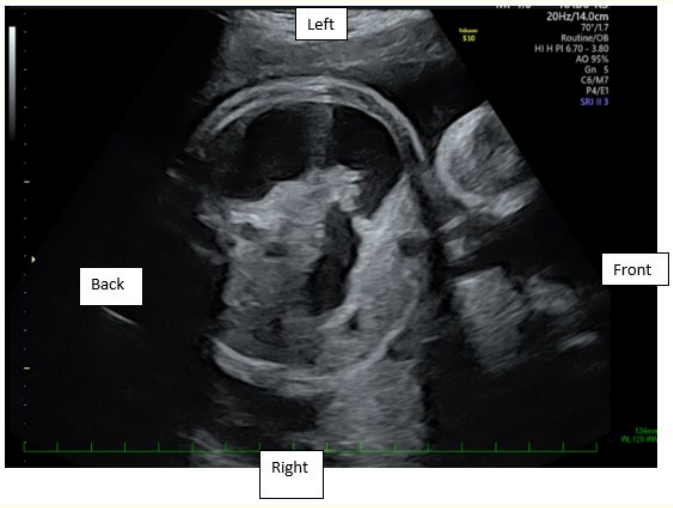

Figure 1: *Obstetric ultrasound image of the fetus showing a double bubble sign in the fetal intestine.*

At 35 weeks and 4 days, the mother went into spontaneous labour and delivered a male baby (1652g). No physical abnormalities were detected. Blood tests and other examinations were performed to screen for abnormalities or complications of prematurity. Abdominal X-ray showed dilated upper small bowel loops, and contrast enema showed an unused microcolon, leading to diagnosis of jejunal atresia in the infant (Figure 2).

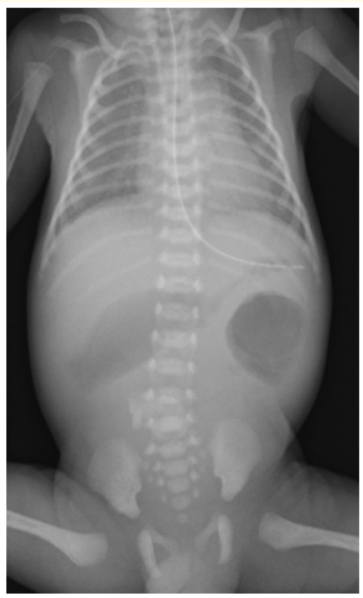

Figure 2: *Supine plain abdominal X-ray on day zero of life, showing a triple bubble sign and a nasogastric tube in-situ.*

The baby underwent surgery on the second day of life. Intraoperative findings included jejunal atresia (type III a) and Meckel's diverticulum. The distal jejunum was twisted over approximately 20 cm, resulting in necrosis of the jejunum (Figure 3a). End-to-back anastomosis of the intestine with resection of the necrotic region and ileal wedge resection of Meckel's diverticulum were performed. A trans-anastomotic tube (TAT) (5 Fr, KangarooTM new enteral feeding tube, Covidien Japan) was placed nasally (Figure 3b). Postoperative temporary disturbed intestinal transit associated with bowel dilation occurred; however, enteral feeding via the TAT could be continued and total parenteral nutrition (TPN) was withdrawn (Figure 4). The baby stayed in the neonatal intensive care unit (NICU) for close monitoring.

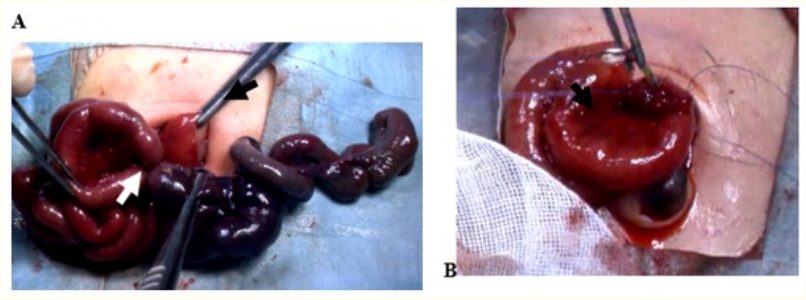

Figure 3: *(a) Proximal jejunum (black arrow) and volvulus of the distal jejunum (white arrow). (b) End-to-back anastomosis, arrow points to the trans-anastomotic tube.*

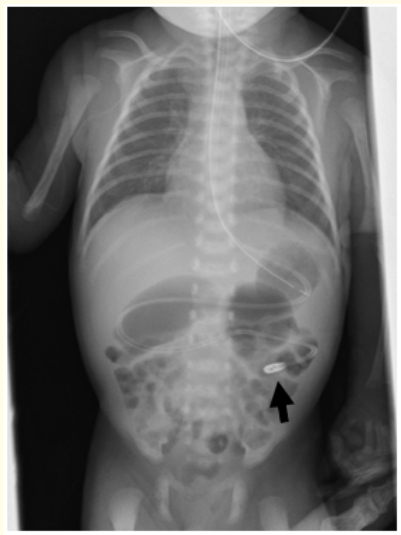

Figure 4: *Postoperative abdominal X-ray showing disrupted intestinal transit with bowel dilation. The arrow points to the trans-anastomotic tube.*

Discussion

Identification of structural abnormalities or chromosomal anomalies in babies during pregnancy is challenging. However, advances in ultrasound equipment have made it possible to visualize anatomical structures of a fetus in increasing detail [3]. The detection rate

varies widely from 10% to 90%, depending on the gestational age at the time of ultrasound examination, site of obstruction, presence of polyhydramnios and other structural abnormalities, degree of bowel dilatation, and experience of the operator [4].

JIA is thought to result from vascular disruption in early gestation. Chromosomal abnormalities are less common in JIA, but it is sometimes associated with other bowel abnormalities (mainly malrotation and gastroschisis), with only a 10% risk of cystic fibrosis and possible occurrence of meconium peritonitis [5]. In our case the fetus had an arm-to-arm inversion of chromosome 5, no previous reports have shown an association between this chromosomal anomaly and jejunal atresia. *De novo* chromosomal abnormalities may cause destruction of genes, and the parents may have a chromosomal rearrangement. The presence of a SUA in our patient can be linked with the anomaly, as SUA has been shown to be associated with chromosomal abnormalities, genetic syndromes, and renal, gastrointestinal, and cardiovascular anomalies [6].

In this case, abnormalities were detected prenatally, the mother was immediately referred to a tertiary level hospital which permitted optimal management. In contrast to our case, in many developing countries intestinal obstruction is detected postnatally. In Tanzania, all pregnant women should attend antenatal clinics for examinations such as routine antenatal ultrasound, but some are unable to access these services due to poverty and lack of sufficient health facilities. Antenatal ultrasound does not focus on assessment of fetal anomalies, and most are detected incidentally. Some facilities do not offer ultrasound tests due to lack of equipment [7] and there are a few sonographers who are trained to detect fetal abnormalities in the prenatal period.

Postoperative care is important in determining the outcome and prognosis of a patient. In a similar case to our case reported from Tanzania, the patient could not receive ideal supportive care due to financial constraints and lack of availability of TPN; therefore, the baby was maintained under a nil per oral protocol and intravenous fluids, which rendered difficulties with nutritional support and electrolyte correction post-surgery, and there was also a lack of NICU care [8]. A better postoperative outcome with TPN can lead to faster recovery and a shorter hospital stay, but financial constraints in another case led to use of maintenance dextrose, normal saline, and lactated Ringers' solution, which led to a poorer outcome compared to use of TPN [9]. These cases suggest that developing countries should examine alternative management approaches that require minimal resources; for example, use of a TAT is very helpful for early enteral nutrition and for shortening the duration of TPN. Studies have shown that a TAT shortens the time to full enteral feeding and significantly reduces the need for central venous access and parenteral nutrition [10].

Conclusion

Prenatal evaluation is crucial for early initial diagnosis of congenital abnormalities and planning for proper management before a baby is born, including early referral to a hospital with appropriate facilities to manage the baby. Developing countries could invest in prenatal diagnosis and screening of fetal anomalies by trainings in detecting these anomalies using tests such as ultrasound, which is affordable, effective, and non-invasive. Genetic tests and genetic counselling for parents on the condition of their baby and the possibility of similar conditions in future pregnancies are also important.

Acknowledgements

The authors would thank all doctors who managed the patients and the mother for her cooperation.

Funding Support

No funding was used for this case report.

Consent

Informed consent was obtained from the mother.

Conflict of Interest

No conflict of interest to disclose.

Authors' Contributions

NO prepared the manuscript, KY, RK and TN were attending doctors. KN, MY and provided expert opinion.

Bibliography

1. Adams SD and Stanton MP. "Malrotation and intestinal atresias". *Early Human Development* 90.12 (2014): 921-925.
2. Edwards L and Hui L. "First and second trimester screening for fetal structural anomalies". *Seminars in Fetal and Neonatal Medicine* 23.2 (2018): 102-111.
3. Hildebrand E. "Prenatal diagnosis of structural malformations and chromosome anomalies: Detection, influence of Body Mass Index and ways to improve screening". Linköping University Electronic Press (2014): 61.
4. Baglaj M., *et al*. "Multiple atresias of the small intestine: A 20- year review". *European Journal of Pediatric Surgery* 18.1 (2008): 13-18.
5. Galani A., *et al*. "Prenatal diagnosis of fetal jejunal atresia: A case report". *Cureus* 13.10 (2021): e18947.
6. Rao R and Platt LD. "Ultrasound screening: Status of markers and efficacy of screening for structural abnormalities". *Seminars in Perinatology* 40.1 (2016): 67-78.
7. Magwesela FM., *et al*. "Pattern of congenital anomalies among pediatric surgical patients in a tertiary care hospital in northern Tanzania". *World Journal of Pediatric Surgery* 5.4 (2022): e000410.
8. Mrindoko P., *et al*. "Ileal atresia in a 10-day-old male baby: A case report". *SAGE Open Medical Case Reports* 9 (2021): 2050313X211042983.
9. Goodluck G., *et al*. "Jejunoileal Atresia in a newborn: Our experience from northern Tanzania and literature review". *Clinical Medicine Insights: Pediatrics* 16 (2022): 11795565221075313.
10. Hall NJ., *et al*. "Trans-anastomotic tubes reduce the need for central venous access and parenteral nutrition in infants with congenital duodenal obstruction". *Pediatric Surgery International* 27.8 (2011): 851-855.

Volume 13 Issue 9 September 2024