International Journal of Environmental Sciences
ISSN: 2229-7359
Vol. 11 No. 8, 2025
https://theaspd.com/index.php

Prenatal Diagnosis Of Duodenal Atresia With Trisomy 21: A Case Report From Indonesia

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Abstract
Introduction: *Duodenal atresia is a leading cause of neonatal intestinal obstruction and is commonly associated with trisomy 21 (Down syndrome). We report a case of duodenal atresia with trisomy 21, a condition rarely documented in Indonesian literature, emphasizing the role of early detection in optimizing outcomes.*
Case Presentation: *A 36-year-old woman ($G_2P_1A_0$) was referred to our tertiary center with polyhydramnios. Prenatal ultrasound revealed the characteristic "double bubble" sign, nasal bone hypoplasia, and hypotelorism. Postnatal genetic testing confirmed trisomy 21. Surgical exploration confirmed an annular pancreas causing the duodenal obstruction, which was successfully corrected with a duodenojejunostomy. The infant had an excellent postoperative recovery.*
Discussion: *Duodenal atresia typically results from failed intestinal recanalization. This case highlights the pivotal role of prenatal ultrasound in identifying not only structural anomalies but also soft markers that guide further genetic investigation. Early diagnosis is particularly critical in lower-middle-income settings, as it facilitates timely referral and multidisciplinary planning, which are key to improving perinatal management.*
Conclusion: *Early prenatal diagnosis of duodenal atresia is a critical trigger for a cascade of interventions including genetic counseling, coordinated delivery planning, and prompt surgical management. This report contributes to the limited published data from Southeast Asia, underscoring the importance of building regional evidence to enhance perinatal care.*
Keyword: *Duodenal Atresia, Trisomy 21, Annular Pancreas, Prenatal Diagnosis, Ultrasound, Indonesia*

INTRODUCTION

Duodenal atresia is a congenital anomaly that represents one of the most frequent causes of neonatal intestinal obstruction, with an estimated incidence ranging from 1 in 2,500 to 10,000 live births. [1,2]. It results from failure of duodenal lumen recanalization during embryogenesis and is commonly detected in the late second or third trimester. Among gastrointestinal anomalies, duodenal atresia is unique due to its strong association with chromosomal abnormalities, particularly trisomy 21 (Down syndrome), which is present in approximately 30–40% of cases [3,4].

Advancements in prenatal ultrasonography have enabled earlier and more accurate diagnosis of duodenal atresia. The characteristic “double bubble” sign is a hallmark sonographic feature indicating proximal duodenal obstruction. When this sign is accompanied by other soft markers such as nasal bone hypoplasia and hypotelorism, the suspicion of trisomy 21 increases, prompting further genetic evaluation [5–8]. Early diagnosis provides a critical window for genetic counseling, delivery planning at a tertiary care center, and perinatal management.

Despite the availability of fetal imaging, published reports on this topic from Southeast Asia, particularly Indonesia, remain limited. This data gap can impact the development of localized management guidelines and patient counseling strategies. Therefore, this report aims not only to highlight the utility of prenatal ultrasound but also to contribute crucial regional data that can inform clinical practice.

Case Presentation

A 36-year-old woman, gravida 2 para 1 abortus 0, at 33 weeks of gestation, was referred to Dr. Hasan Sadikin General Hospital, Bandung, due to polyhydramnios. Our detailed ultrasound evaluation confirmed a single live intrauterine fetus and identified several key findings. A transverse abdominal scan revealed the classic “double bubble” sign, confirming duodenal obstruction (Figure 1). Further craniofacial assessment demonstrated hypotelorism and nasal bone hypoplasia, which significantly raised the suspicion for an associated aneuploidy, specifically trisomy 21 (Figure 2). Fetal echocardiography and Doppler studies were within normal limits.

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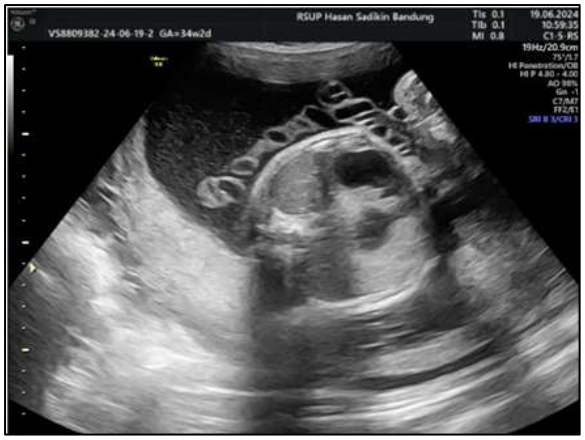

Figure 1. Transverse abdominal section showing "Double Bubble" sign

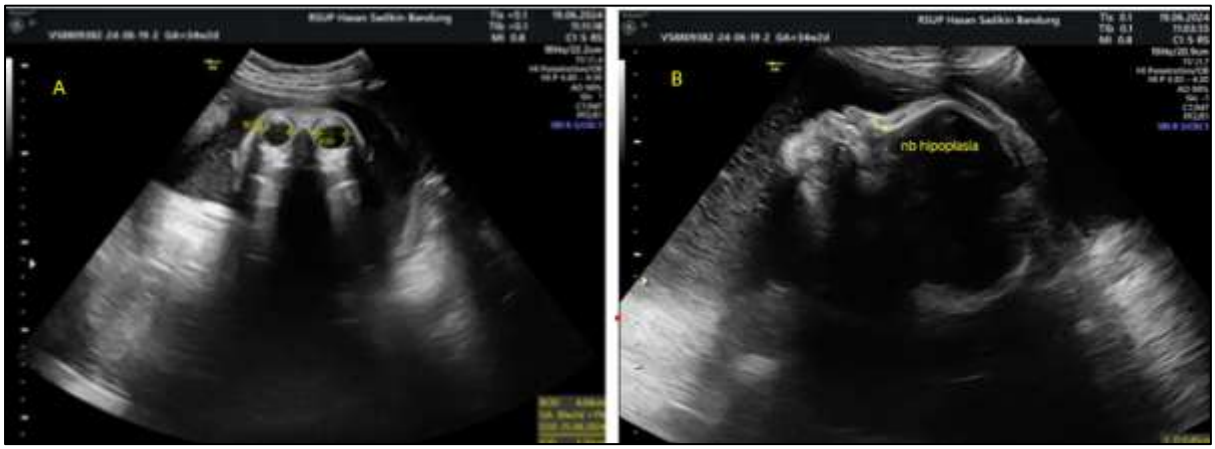

Figure 2. Facial profile assessment showing Hypotelorism (A) and Nasal bone hypoplasia (B)

Delivery was performed via cesarean section at 37 weeks of gestation. A live male infant was born weighing 1990 grams, with a length of 43 cm and Apgar scores of 8 and 9 at 1 and 5 minutes, respectively. Postnatal abdominal radiography confirmed duodenal obstruction. Surgical exploration revealed gastric distension and narrowing of the duodenum caused by an annular pancreas encircling the first and second portions of the duodenum (Figure 3). The infant underwent successful duodenojejunostomy performed by a pediatric surgeon. Chromosomal analysis confirmed a male karyotype with trisomy 21 (Figure 4).

Figure 3. Annular pancreas encircling the first part of the duodenum

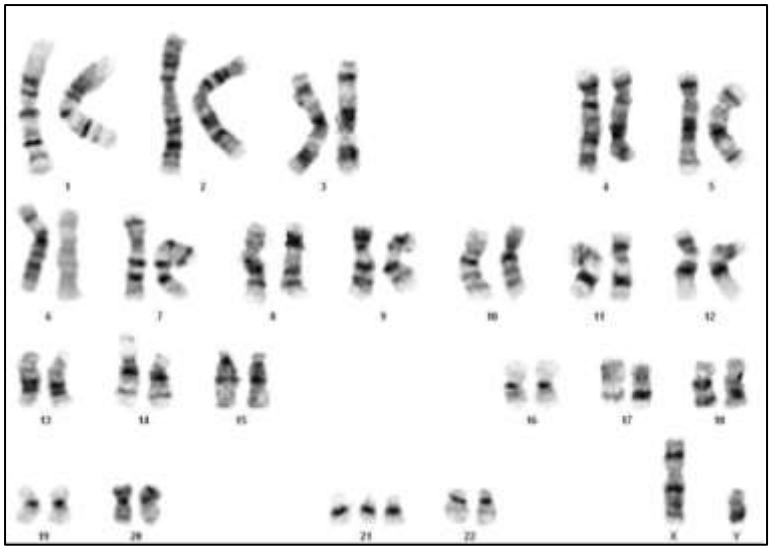

Figure 4. Chromosome analysis result showing trisomy 21

DISCUSSION

Duodenal atresia arises from failure of intestinal recanalization during the 8th to 10th weeks of gestation [2,3]. It is classified into three types: type I (duodenal web), type II (fibrous cord connecting proximal and distal segments), and type III (complete separation with a V-shaped mesenteric defect). In this case, the obstruction was caused by annular pancreas, a rare extrinsic etiology of duodenal obstruction [3,4].

Prenatal ultrasound plays a pivotal role in identifying gastrointestinal anomalies. The "double bubble" sign is a reliable indicator of duodenal atresia and can be visualized in the late second or third trimester [6]. This finding, when accompanied by polyhydramnios and other soft markers such as nasal bone hypoplasia and hypotelorism, raises the index of suspicion for trisomy 21 [7,8]. Our patient presented with these features, and trisomy 21 was confirmed by chromosomal analysis postnatally.

Duodenal atresia is associated with trisomy 21 in approximately 30–40% of cases, making genetic counseling and karyotyping essential when suspected prenatally [5,9]. Early diagnosis allows for planned delivery in a tertiary center, multidisciplinary coordination, and timely surgical intervention. Definitive treatment is surgical, with either duodeno-duodenostomy or duodenojejunostomy, and outcomes are generally favorable with current neonatal care standards [10–12].

The management of this particular case at our institution serves as a functional model for resource-limited settings. The crucial step was the detailed initial ultrasound, which triggered a cascade of well-coordinated actions: multidisciplinary consultation (obstetrics, pediatrics, surgery), informed parental counseling, and planned delivery at a tertiary center equipped for neonatal surgery. This proactive pathway, initiated prenatally, was instrumental in achieving a favorable outcome and demonstrates that high standards of perinatal care are attainable.

CONCLUSION

Prenatal ultrasound in cases of suspected duodenal atresia should be viewed not merely as a diagnostic tool, but as a critical catalyst for a coordinated, multidisciplinary management pathway. This case demonstrates that a detailed sonographic evaluation, encompassing both structural anomalies and aneuploidy markers, is the foundational step toward optimizing neonatal outcomes. This report represents one of the few published cases from Indonesia describing prenatal diagnosis of duodenal atresia associated with trisomy 21, emphasizing the importance of building regional evidence for better perinatal care.

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