ISSN: 2584-0355
DOI: doi.org/10.51219/MCCRJ/Mohammed-Ech-Chebab/466

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Medical & Clinical Case Reports Journal

https://urfpublishers.com/journal/case-reports

Vol: 4 & Iss: 2

Case Report

Duodenal Atresia in a Neonate with Down Syndrome Presenting with Bilious Vomiting

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Citation: Ech-Chebab M, Ayyad A, Messaoudi S, Amrani R. Duodenal Atresia in a Neonate with Down Syndrome Presenting with Bilious Vomiting. *Medi Clin Case Rep J* 2026;4(2):1710-1712. DOI: doi.org/10.51219/MCCRJ/Mohammed-Ech-Chebab/466

Received: 22 April, 2026; **Accepted:** 25 April, 2026; **Published:** 27 April, 2026

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A B S T R A C T

Background: Duodenal atresia is a congenital obstruction of the duodenum and a well-known cause of neonatal intestinal obstruction. It is frequently associated with Down syndrome.

Case presentation: We report the case of a term female neonate admitted on day 8 of life for bilious vomiting and dehydration. Clinical examination revealed dysmorphic features suggestive of Down syndrome, hypotonia, and a non-distended abdomen. Abdominal radiography demonstrated the characteristic "double bubble" sign with absence of distal bowel gas. Surgical exploration confirmed intrinsic duodenal atresia at the level of the second portion of the duodenum. A duodeno-duodenostomy was performed with favorable postoperative outcome. Echocardiography identified a small atrial septal defect, and karyotype confirmed trisomy 21.

Conclusion: Duodenal atresia should be suspected in any neonate presenting with bilious vomiting, particularly in those with Down syndrome. Early diagnosis and prompt surgical management are essential to improve outcomes.

Keywords: Neonate; Duodenal atresia; Down syndrome; Bilious vomiting; Double bubble

Introduction

Duodenal atresia is a congenital anomaly resulting from failure of duodenal recanalization during early embryogenesis. It represents one of the most common causes of neonatal intestinal obstruction, with an estimated incidence ranging from 1 in 5,000 to 1 in 10,000 live births[1,2].

This condition is frequently associated with other congenital anomalies, particularly Down syndrome, which is reported in approximately 20-30% of cases[3]. Congenital heart defects are also commonly associated and may significantly influence prognosis[4].

Clinically, duodenal atresia typically presents in the neonatal period with bilious vomiting and minimal abdominal distension. The diagnosis is often suggested by abdominal radiography demonstrating the characteristic "double bubble" sign, which is considered pathognomonic[5].

Early recognition is essential to prevent complications and allow timely surgical management. We report a case of duodenal atresia in a neonate with Down syndrome revealed by neonatal intestinal obstruction.

Case Presentation

We report the case of a female neonate born at term to a 21-year-old mother with an uneventful pregnancy. The parents were non-consanguineous. The delivery was vaginal, and the birth weight was 2.4 kg.

The newborn was referred to our neonatal intensive care unit on day 8 of life for bilious vomiting associated with dehydration and delayed passage of meconium.

On physical examination, the neonate presented dysmorphic features suggestive of Down syndrome, including characteristic facial appearance and hypotonia. The infant was dehydrated, with a soft, non-distended abdomen. Cardiac auscultation revealed a systolic murmur.

Abdominal radiography demonstrated a typical "double bubble" sign with absence of distal bowel gas, consistent with a proximal intestinal obstruction **(Figure 1)**.

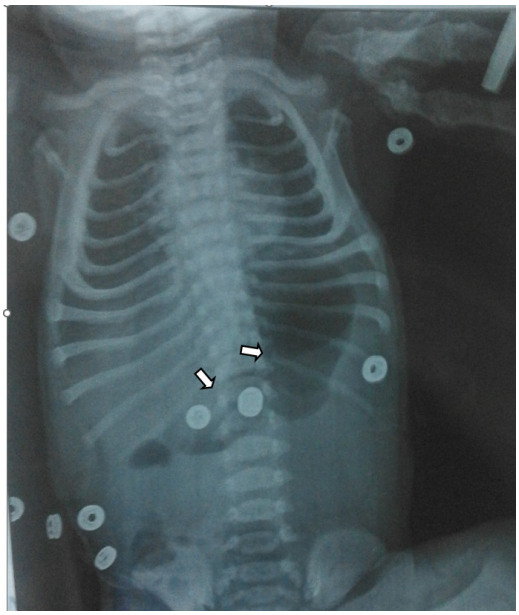

Figure 1: Abdominal radiograph showing the classic "double bubble" sign (arrows), with gastric and proximal duodenal dilation and absence of distal bowel gas, consistent with duodenal atresia

Surgical exploration confirmed intrinsic duodenal atresia located at the second portion of the duodenum, with significant proximal dilation.

A duodeno-duodenostomy was performed. The postoperative course was favourable, with resolution of bilious vomiting within four days.

Further investigations revealed an atrial septal defect on echocardiography, and karyotype analysis confirmed trisomy 21.

At follow-up, the patient showed good clinical evolution and is currently under regular cardiology supervision.

Discussion

Duodenal atresia is a well-recognized cause of neonatal intestinal obstruction, with an estimated incidence ranging from 1 in 5,000 to 1 in 10,000 live births[1,2]. It results from failure of recanalization of the duodenum during embryogenesis and most commonly involves the second portion of the duodenum[6].

A strong association exists between duodenal atresia and chromosomal abnormalities, particularly Down syndrome, reported in approximately 20-30% of cases[3,7]. In addition, associated congenital anomalies are frequent, especially cardiac malformations, which may affect prognosis and perioperative management[4,8].

The classical clinical presentation includes early-onset bilious vomiting with minimal or absent abdominal distension, reflecting a high intestinal obstruction[5,9]. However, delayed presentation, as observed in our case, may occur and can lead to diagnostic delay, particularly in low-resource settings[10].

Radiologically, the "double bubble" sign on abdominal radiography is considered pathognomonic of duodenal obstruction and allows rapid diagnosis without the need for further imaging in most cases[5,11].

In all cases of duodenal atresia, a systematic search for associated anomalies is essential. Echocardiography is particularly important given the high frequency of congenital heart disease, as observed in our patient who presented with an atrial septal defect[4,12].

Surgical treatment is the standard of care and consists of duodeno-duodenostomy, which provides excellent outcomes when performed early[13]. Advances in neonatal care and surgical techniques have significantly improved survival rates, which now exceed 90% in developed settings[14].

Our case highlights the importance of considering duodenal atresia in any neonate presenting with bilious vomiting, especially in the presence of Down syndrome. Early diagnosis and prompt management are essential to prevent complications and improve prognosis.

Conclusion

Duodenal atresia is a common cause of neonatal intestinal obstruction and is frequently associated with Down syndrome. The presence of bilious vomiting in a neonate, particularly without significant abdominal distension, should raise strong suspicion for a proximal intestinal obstruction.

Early diagnosis based on clinical presentation and radiological findings, followed by prompt surgical management, is essential to ensure favorable outcomes. Systematic screening for associated anomalies, especially cardiac defects, remains crucial in the overall management of these patients.

Disclosures

Conflicts of interest

The authors declare no conflicts of interest.

Patient consent

Written informed consent was obtained from the patient's parents for publication of this case and accompanying images.

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