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Original Article

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Clinical outcomes after Kasai portoenterostomy in biliary atresia: role of early surgery and liver surface telangiectasia in a resource-limited setting

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ABSTRACT

Aims: Biliary atresia (BA) remains a leading cause of pediatric liver failure worldwide, with outcomes heavily influenced by the timing of intervention and healthcare resources. In low- and middle-income countries, delayed diagnosis and limited access to specialized care contribute to poorer outcomes following Kasai portoenterostomy (KPE). This study aimed to evaluate clinical outcomes and identify prognostic factors in infants undergoing KPE at a tertiary pediatric center.

Methods: This retrospective observational study included all consecutive infants with BA who underwent KPE at our center between January 2022 and December 2025. Data on demographic characteristics, preoperative biochemical parameters, intraoperative findings, and postoperative outcomes were collected. Survival and jaundice clearance were assessed at 3, 6, and 12 months. Associations between clinical variables and outcomes were analyzed using the Mann–Whitney U test, the Chi-square test, or Fisher's exact test, with $p<0.05$ considered statistically significant.

Results: A total of 52 infants were included, with a median age at operation of 71 days. Liver surface telangiectasia was present in 63.5% of patients. The 30-day mortality was 7.7%, and survival rates at 3 months, 6 months, and 1 year were 65.4%, 51.9%, and 48.1%, respectively. Jaundice clearance at 6 months was achieved in 34.6% of patients. Younger age at operation was significantly associated with jaundice clearance (median 62 vs 72 days, $p=0.046$). Preoperative direct bilirubin was significantly higher among non-survivors ($p=0.028$). Liver surface telangiectasia showed a trend toward poorer jaundice clearance ($p=0.055$). Kaplan–Meier analysis demonstrated a progressive decline in survival, particularly within the early postoperative period.

Conclusion: Outcomes of BA following KPE remain suboptimal in this setting, largely due to delayed presentation and advanced disease at surgery. Early surgical intervention is the most important modifiable factor associated with improved bile drainage, while elevated direct bilirubin and liver surface telangiectasia may indicate poorer prognosis. Strengthening early diagnosis and referral pathways is essential to improving outcomes in resource-limited settings.

Keywords: Biliary atresia, Kasai portoenterostomy, jaundice clearance, survival analysis, liver fibrosis, pediatric surgery

INTRODUCTION

Biliary atresia (BA) is a progressive fibro-obliterative cholangiopathy of infancy characterized by inflammation and obliteration of the extrahepatic biliary tree, leading to persistent cholestasis, biliary cirrhosis, and ultimately liver failure if untreated. It represents the most common indication for pediatric liver transplantation worldwide and continues to pose significant clinical and research challenges despite advances in diagnosis and management.[1]

In low- and middle-income countries (LMICs), the management of BA faces several unique challenges, including delayed diagnosis, limited awareness among primary healthcare providers, restricted access to specialized hepatobiliary surgical services, and limited availability of pediatric liver transplantation. These factors often result in late presentation and reduced success rates following Kasai portoenterostomy (KPE). Studies from several LMICs have demonstrated that many infants undergo surgery beyond the optimal age for intervention, which significantly affects postoperative bile drainage and long-term native liver survival.[2]

While Kasai Porto enterostomy is the only hope for these patients in LMICs, informed decision-making is sometimes challenging in the pre- and post-operative period. Aside from

advanced age, biochemical markers, and histology, there is no universally accepted gross liver finding that can predict poorer outcome or advanced liver damage in these patients. Zhang et al.[3] reported that the hepatic subcapsular telangiectasia sign was present in all infants with BA and absent in non-BA cholestatic disorders, with 100% sensitivity and specificity for diagnosis; however, they also noted that this could be seen even in very young infants without obvious cirrhosis, indicating that the sign is not simply a marker of advanced fibrosis.

Despite these challenges, evidence suggests that meaningful clinical benefit can still be achieved when KPE is performed in specialized centers with appropriate surgical expertise and postoperative care. Evaluating institutional outcomes in LMIC settings is therefore essential for identifying gaps in care, improving referral pathways, and optimizing management strategies for children with BA.[2,4]

The objective of this study was to evaluate the clinical outcomes of infants with BA undergoing KPE at a tertiary pediatric center. In addition, the study aims to analyze postoperative outcomes and identify age at surgery and liver surface telangiectasia as prognostic factors following KPE.

METHODS

The study was conducted in accordance with the ethical principles set forth in the Declaration of Helsinki. Ethical approval was obtained from the Ethics Review Board of the Bangladesh Shishu Hospital and Institute (Date: 15.01.2026, Decision No: Admin/BSHI/2026/207). Patient confidentiality was maintained throughout the study, and all data were anonymized prior to analysis.

This retrospective observational study was conducted at a tertiary pediatric referral center providing specialized pediatric surgical services. The study evaluated the clinical outcomes of infants diagnosed with BA who underwent KPE at this institution.

All consecutive infants diagnosed with BA who underwent KPE by the same surgery team between January 2022 and December 2025 were included in the study. Patients with incomplete clinical data and those who did not undergo KPE (including patients who were not fit for surgery and parents who refused surgery) were excluded.

Data were collected retrospectively from hospital medical records, operative notes, and follow-up documentation using a structured data collection form. The following variables were recorded: age at surgery, preoperative biochemical parameters, intraoperative findings (including liver surface telangiectasia), postoperative biochemical parameters, jaundice clearance, and survival outcomes. Liver surface telangiectasia was defined as the macroscopic presence of subcapsular vessels on the liver surface.

Statistical Analysis

Primary outcomes were survival with native liver and jaundice clearance at 3, 6, and 12 months. Serum total bilirubin below 2.0 mg/dl at 6 months after surgery was considered jaundice clearance. Continuous variables were expressed as mean or median (range), and categorical variables as frequencies and percentages. Comparisons were performed using the Mann–Whitney U test, Chi-square test, or Fisher's exact test as appropriate. A p-value <0.05 was considered statistically significant.

RESULTS

A total of 52 infants with BA who underwent KPE during the study period were included in the analysis. **Table 1** summarizes the independent variables. The mean age at operation was 78.3 days, with a median of 71 days (range 33–155 days). Liver surface telangiectasia (**Figure 1**) was present in 33 patients (63.5%).

Table 1. Comparison of variables between survivors and non-survivors

Variable	Survivors (n=25)	Non-survivors (n=27)	p-value
Male sex, n (%)	14 (56.0%)	18 (66.7%)	0.614
Age at operation (days), median (range)	68 (33–155)	72 (33–155)	0.077
Preoperative total bilirubin (mg/dl), mean	12.2	12.9	0.667
Preoperative direct bilirubin (mg/dl), mean	7.86	9.69	0.028
Liver surface telangiectasia present, n (%)	10 (40.0%)	19 (70.4%)	0.027

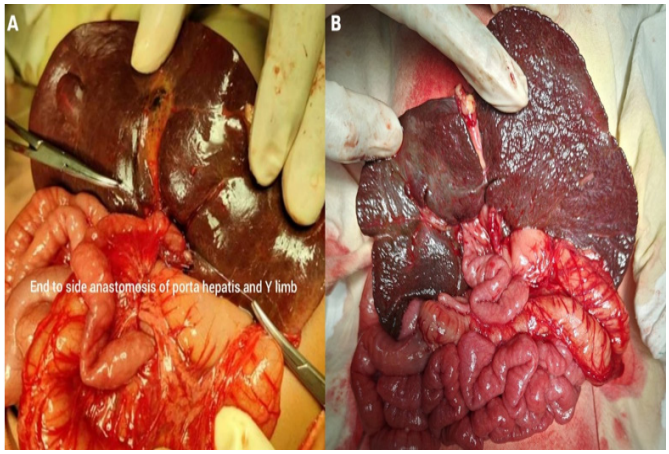

Figure 1. A- Liver without surface telangiectasia; **B-** Liver with surface telangiectasia

The 30-day mortality was 7.7% (4/52). These patients were categorized as non-clearance of jaundice. The mean preoperative total bilirubin was 12.6 mg/dl, decreasing to 7.8 mg/dl at 2 weeks after surgery among 48 patients (excluding 4 patients with 30-day mortality). Mean direct bilirubin decreased from 8.8 mg/dl preoperatively to 5.4 mg/dl at two weeks. At three months, mean total bilirubin decreased further to 5.1 mg/dl, with a mean direct bilirubin of 4.0 mg/dl, demonstrating progressive biochemical improvement among surviving patients. **Table 2** summarizes the survival outcomes and jaundice clearance.

Those who achieved jaundice clearance underwent surgery at a significantly younger age than those who did not. The median age at operation was 62 days in the jaundice-clearance group, compared with 72 days in the non-clearance group

Table 2. Survival outcomes and jaundice clearance

Outcome	Value
30-day mortality	4/52 (7.7%)
3-month survival	34/52 (65.4%)
6-month survival	27/52 (51.9%)
1-year survival	25/52 (48.1%)
Jaundice clearance at 6 months	18/52 (34.6%)

(p=0.046). There was no significant difference in preoperative total bilirubin (12.34 vs 12.50 mg/dl, p=0.942) or preoperative direct bilirubin (8.06 vs 8.14 mg/dl, p=0.866) between the two groups.

Liver surface telangiectasia showed a borderline association with jaundice clearance. Jaundice clearance occurred in 11 of 13 patients (84.6%) without telangiectasia compared with 7 of 15 patients (46.7%) with telangiectasia, p=0.055 (**Table 3**).

Table 3. Factors associated with jaundice clearance

Variable	Cleared (mean/median)	Not cleared (mean/median)	p-value
Age at operation (days)	62.5/62	88.6/72	0.046
Total bilirubin (mg/dl)	12.34/12.5	12.50/13	0.942
Direct bilirubin (mg/dl)	8.06/8.5	8.14/7.5	0.866
Telangiectasia (yes vs no)	7/15 vs 11/13	-	0.055

Multivariable Logistic Regression for Jaundice Clearance

A multivariable logistic regression model was performed, including age at operation, preoperative direct bilirubin, and liver surface telangiectasia. None of the variables remained independently associated with jaundice clearance in the adjusted model (**Table 4**).

Table 4. Regression analysis

Variable	OR	95% CI	p-value
Age at operation (per day)	1.015	0.954–1.081	0.631
Preoperative direct bilirubin (mg/dl)	0.585	0.262–1.308	0.191
Liver surface telangiectasia	0.23	0.03–2.05	1.000

OR: Odds ratio, CI: Confidence interval

Kaplan–Meier analysis demonstrating overall survival of children with BA following KPE over a 12-month follow-up period. Survival probability declined progressively during the first year, with the greatest reduction occurring in the early postoperative months (**Figure 2**).

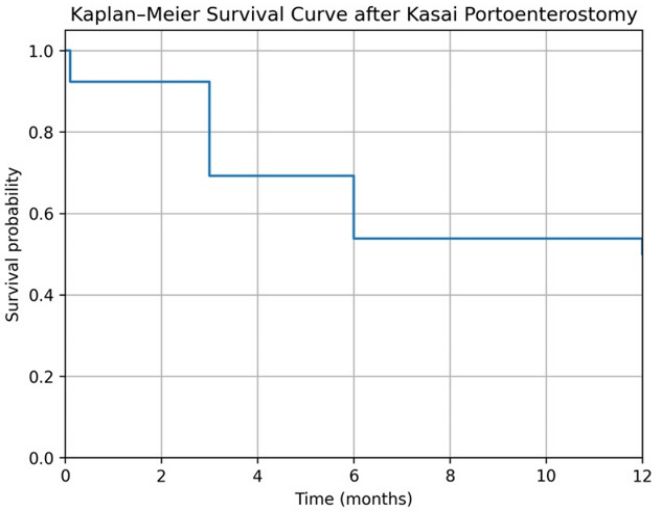

Figure 2. Kaplan–Meier survival analysis demonstrating overall survival of children with biliary atresia following Kasai portoenterostomy over a 12-month follow-up period

DISCUSSION

This study evaluates the clinical outcomes and prognostic factors following KPE in a cohort of infants with BA from a tertiary pediatric center in Bangladesh. The findings highlight the impact of early surgical intervention, biochemical parameters, and intraoperative findings on postoperative outcomes in a resource-limited setting.

The overall one-year survival with native liver in this cohort was 48.1%, which is lower than outcomes reported from high-income countries, where survival rates often exceed 60-70% following centralized care and early intervention.[4,9] However, the outcomes observed in this study are comparable to reports from other LMICs, where delayed presentation and limited access to specialized care remain significant challenges.[2,5,10,11] This reinforces the persistent disparity in outcomes for BA between high-resource and low-resource settings.

Age at surgery emerged as an important determinant of postoperative outcome, particularly in relation to jaundice clearance. Patients who achieved jaundice clearance underwent surgery at a significantly younger age compared to those who did not. This finding is consistent with multiple studies demonstrating that early KPE—ideally before 60 days of life—is associated with improved bile drainage and long-term native liver survival.[4,9] Although age did not show a statistically significant association with survival in this cohort, the observed trend likely reflects the limited sample size and reduced statistical power.

Preoperative direct bilirubin was significantly higher among non-survivors, suggesting that greater cholestatic burden at presentation may reflect more advanced disease and poorer hepatic reserve. Similar associations have been reported in previous studies, where elevated bilirubin levels correlate with worse postoperative outcomes and increased risk of liver failure.[6,8] However, total bilirubin did not show a significant association, indicating that direct bilirubin may be a more sensitive marker of disease severity in this context.

A key and innovative aspect of this study is assessing liver surface telangiectasia as a potential prognostic marker. Although initially identified as a diagnostic indicator of BA with high sensitivity and specificity,[3] its prognostic value remains uncertain. Although liver surface telangiectasia was associated with poorer outcomes on univariable analysis, its effect could not be reliably estimated in adjusted modeling due to sparse data and model instability. This likely reflects

the small sample size and quasi-separation, rather than the absence of a true association. The observed trend is clinically important and warrants further study.

The rate of jaundice clearance at six months in this cohort was 34.6%, which is lower than rates reported in high-income settings but comparable to those reported in other LMIC studies.[10-13] Jaundice clearance is a key early surrogate marker of successful biliary drainage and has been consistently associated with improved long-term outcomes, including native liver survival.[7,12] The relatively low clearance rate observed in this study may reflect delayed presentation, advanced disease at the time of surgery, and limitations in postoperative care.

The Kaplan–Meier survival analysis demonstrated a progressive decline in survival during the first year, with the most significant drop occurring in the early postoperative period. This pattern is consistent with previous reports, where early postoperative mortality is often related to sepsis, cholangitis, or liver failure.[2,6] The absence of loss to follow-up in this study strengthens the validity of the survival analysis and enhances the reliability of outcome assessment.

Overall, the findings of this study emphasize the critical importance of early diagnosis, timely referral, and surgical intervention in improving outcomes for BA in LMIC settings. In addition, the potential role of intraoperative findings, such as liver surface telangiectasia, as prognostic indicators warrant further research.

Limitations

The absence of histopathological correlation of the liver surface telangiectasia restricted deeper evaluation of disease severity, and long-term outcomes beyond one year could not be assessed. Additionally, as a tertiary referral center, we may be subject to referral bias, with a higher proportion of advanced cases included.

Recommendations

Despite these limitations, the study provides valuable insights into outcomes in BA in a low-resource setting. Future efforts should focus on establishing national screening and early referral programs to facilitate timely diagnosis and intervention. Increasing awareness among primary healthcare providers, developing centralized management pathways, and promoting multicenter collaborative studies are essential to improve outcomes. Further research is also warranted to explore the prognostic significance of intraoperative findings such as liver surface telangiectasia and to optimize postoperative care strategies.

CONCLUSION

BA continues to be associated with suboptimal outcomes in low-resource settings, primarily due to delayed presentation and advanced disease at the time of surgery. Early age at KPE remains the most important modifiable factor associated with improved bile drainage, while elevated preoperative direct bilirubin and the presence of liver surface telangiectasia may indicate more advanced disease and poorer prognosis. Strengthening early detection strategies and referral pathways is essential to improve clinical outcomes in this population.

ETHICAL DECLARATIONS

Ethics Committee Approval

Ethical approval was obtained from the Ethics Review Board of the Bangladesh Shishu Hospital and Institute (Date: 15.01.2026, Decision No: Admin/BSHI/2026/207).

Informed Consent

As this was a retrospective study, formal written informed consent was not required and was therefore not obtained.

Peer Review Process

This manuscript was subject to external peer review.

Conflict of Interest

The authors declare no conflicts of interest related to this study.

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Author Contributions

Concept: MSH; Design: MSH; Control: AR; Resources: MSH; UFKBB, RM, Materials: MSH, UFKB; Data Collection and/or Processing: UFKBB, RM; MSH; Analysis and/or Interpretation: MSH, SMNI; Literature Review: MSH, RM; Writing the Article: MSH; Critical Review: MSH; UFKBB, RM, SMNI, AR.

REFERENCES

1. Bezerra JA, Wells RG, Mack CL, et al. Biliary atresia: clinical and research challenges for the twenty-first century. *Hepatology.* 2018;68(3):1163-1173. doi:10.1002/hep.29905
2. Hong KW, Igiraneza D, Davis J, et al. Should we offer surgery for biliary atresia in low-resource settings? *J Surg Res.* 2021;257:419-424. doi:10.1016/j.jss.2020.08.004
3. Zhang K, Tang Y, Liu R, et al. Intraoperative hepatic subcapsular spider-like telangiectasia sign for the definitive diagnosis of biliary atresia. *BMC Pediatr.* 2023;23(1):63. doi:10.1186/s12887-022-03831-z
4. Davenport M, Ong E, Sharif K, et al. Biliary atresia in England and Wales: results of centralization and new benchmark. *J Pediatr Surg.* 2011;46(9):1689-1694. doi:10.1016/j.jpedsurg.2011.04.013
5. Masturina M, Prihaningtyas RA, Setyoboedi B, Arief S. Late referral of biliary atresia and poor clinical outcomes. *Korean J Gastroenterol.* 2025;85(3):382-388. doi:10.4166/kjg.2025.005
6. DeRusso PA, Ye W, Shepherd R, et al. Growth failure and outcomes in infants with biliary atresia: report from the Biliary Atresia Research Consortium. *Hepatology.* 2007;46(5):1632-1638. doi:10.1002/hep.21923
7. Lee WS, Ong SY, Foo HW, et al. Chronic liver disease is universal in children with biliary atresia living with native liver. *World J Gastroenterol.* 2017;23(43):7776-7784. doi:10.3748/wjg.v23.i43.7776
8. Khayat A, Alamri AM, Saadah OI. Outcomes of late Kasai portoenterostomy in biliary atresia: a single-center experience. *J Int Med Res.* 2021;49(5):3000605211012596. doi:10.1177/03000605211012596
9. Davenport M, Makin E, Ong E, et al. The outcome of a centralization program in biliary atresia: twenty years and beyond. *Ann Surg.* 2025;281(4):608-614. doi:10.1097/SLA.0000000000006273
10. Al-Hussaini A, Abanemai M, Alhebbi H, et al. The epidemiology and outcome of biliary atresia: Saudi Arabian national study. *Front Pediatr.* 2022;10:921948. doi:10.3389/fped.2022.921948

11. Holdar S, Alsaleem B, Asery A, Al-Hussaini A. Outcome of biliary atresia among Saudi children. *Saudi J Gastroenterol.* 2019;25(3):176-180. doi:10.4103/sjg.SJG_306_18
12. Sadiq J, Lloyd C, Hodson J, et al. Long-term clinical and socioeconomic outcomes of children with biliary atresia. *JGH Open.* 2023;7(12):841-847. doi:10.1002/jgh3.12980
13. Alaifan MA, Simbawa SH, Fayoumi TA, et al. Outcomes of biliary atresia in a single center in Jeddah. *Cureus.* 2022;14(8):e27871. doi:10.7759/cureus.27871
14. Vats A, Bhatia A, Kumar Y. Biliary atresia: a meta-analysis of Indian studies. *J Clin Exp Hepatol.* 2026;16(1):103156. doi:10.1016/j.jceh.2025.103156

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Original Article

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Comparison of short-chain fatty acid (SCFA) patterns in children with and without Hirschsprung's disease

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ABSTRACT

Aims: Hirschsprung's disease (HD) is a congenital gastrointestinal anomaly that causes morbidity and increases the risk of mortality. The increased risk of infection in HD is related to several factors, including dysbiosis of the intestinal microbiota, which plays an important role in maintaining intestinal function through the production of short-chain fatty acids (SCFAs). This study aimed to determine the differences in SCFA levels in children with HD and children without HD.

Methods: This study used an observational analytic cross-sectional design. The subjects were pediatric patients treated at Cipto Mangunkusomo Hospital from January to June 2024 who met the inclusion and exclusion criteria. A total of 17 pediatric patients were included, consisting of 9 children with HD and 8 children without HD. Fecal SCFA levels were analyzed and compared between the two groups.

Results: SCFA levels in children with HD (2.4±1.6) were significantly lower than in children without HD (7.9±2.4) ($p<0.05$). There were no statistically significant differences ($p>0.05$) between the HD group (n=9) and the non-HD group (n=8) in individual SCFA components, including butyrate, propionate, valerate, and acetate levels.

Conclusion: Children with HD have significantly lower total fecal SCFA levels compared to children without HD, while no significant differences were found in individual SCFA components.

Keywords: Hirschsprung's disease, short-chain fatty acids, Hirschsprung-associated enterocolitis

INTRODUCTION

Hirschsprung's disease (HD) is a congenital gastrointestinal disorder caused by the absence of ganglion cells in the submucosal and myenteric plexuses, resulting in loss of normal intestinal peristalsis and functional bowel obstruction.[1] The incidence of HD has been reported to range from approximately 1 in 5,000 to 1 in 10,000 live births.[2] This condition is associated with substantial morbidity and a risk of mortality, particularly due to complications such as Hirschsprung-associated enterocolitis (HAEC), which remains a life-threatening condition in affected patients.[3,4] HAEC may occur in 6-60% of patients before definitive surgery and in 25-37% after surgery, with reported mortality rates reaching up to 10%.[3-5]

The pathophysiology of HAEC has not been fully elucidated. Several factors have been implicated, including impaired immune responses, dysfunction of the epithelial barrier, and intestinal dysbiosis.[6] The intestinal microbiota plays an essential role in maintaining gut homeostasis, and one of its key functions is the production of short-chain fatty acids (SCFAs).[7] SCFAs, mainly acetate, propionate, and butyrate, account for approximately 90-95% of SCFAs in the colon and are produced through bacterial fermentation of non-digestible carbohydrates.[7,8] These metabolites serve as an important energy source for colonocytes, contribute to epithelial barrier integrity, and exert anti-inflammatory and immunomodulatory effects.[8,9]

Alterations in SCFA production have been associated with various gastrointestinal and inflammatory conditions. A prospective study by Rao et al.[10] showed that neonates with congenital gastrointestinal anomalies had lower fecal SCFA concentrations compared with healthy infants. Decreased SCFA levels may increase the risk of colonic infection and impair mucosal defense.[8] Demehri et al.[3] reported up to a fourfold reduction in fecal SCFA concentrations in patients

with HD (particularly those with a history of HAEC), supporting the role of dysbiosis and altered microbial metabolism in the pathogenesis of enterocolitis.

Most previous studies remain limited. SCFA concentrations in early life are known to vary with age and dietary factors, with considerable variability during the first year of life.[11] This variability complicates the interpretation of SCFA profiles across different pediatric populations. This study aimed to compare fecal SCFA levels in children with HD and children without HD. By evaluating total SCFA levels and individual SCFA components, this study seeks to provide further insight into the role of altered microbial metabolism in HD and its potential implications for intestinal health and susceptibility to complications. Data on SCFA profiles in children with HD remain limited, particularly in developing countries, highlighting the need for further investigation.

METHODS

This study was conducted with approval from the Ethics Committee of Faculty of Medicine, Universitas Indonesia (Date: 18.12.2023, Decision No: KET-1814/UN2.F1/ETIK/PPM.00.02/2023). The study was performed in accordance with the principles of the Declaration of Helsinki. Written informed consent was obtained from the parents or legal guardians of all participants prior to enrollment in the study. Patient confidentiality and data anonymity were maintained throughout the study.

This study was an observational analytic study with a cross-sectional design. The study was conducted at Cipto Mangunkusumo Hospital, Jakarta, Indonesia, from January to June 2024. The target population was pediatric patients with HD, and the accessible population consisted of pediatric patients treated at Cipto Mangunkusumo Hospital during the study period.

Subjects were recruited using a consecutive sampling method. A total of 17 pediatric subjects were included in this study, consisting of 9 children with HD and 8 children without HD. The control group consisted of pediatric patients without HD who were treated at Cipto Mangunkusomo Hospital during the study period and fulfilled the inclusion and exclusion criteria. Stool samples for SCFA analysis were collected as part of the study protocol after informed consent had been obtained from parents or legal guardians. The cost of SCFA examination was covered by the research project.

The inclusion criteria were: pediatric patients aged 1 month to 18 years with HD, either before or after definitive surgery (within 28 days postoperatively); and pediatric patients aged 1 month to 18 years without HD or other congenital gastrointestinal diseases. The exclusion criteria were: Patients with other congenital gastrointestinal anomalies; and patients with a history of antibiotic or probiotic use within one month prior to stool sample collection, including patients with recent perioperative antibiotic exposure within that period. Due to the limited number of eligible patients during the study period, a formal sample size calculation was not performed, and the study should be considered exploratory in nature.

Fecal SCFA concentrations were analyzed using gas chromatography–mass spectrometry (GC-MS) at the Laboratorium Prodia Kramat. Stool samples were collected in sterile containers and stored at 4°C until analysis. Prior to analysis, the samples were prepared according to the laboratory protocol, including homogenization, dilution, centrifugation, and derivatization. Quantification of acetate, propionate, butyrate, and valerate was performed using standard calibration curves with internal standards. Quality control procedures were applied throughout the analysis to ensure measurement accuracy and reproducibility.

Statistical Analysis

Data were analyzed using statistical software. Continuous variables were presented as mean±standard deviation for normally distributed data and as median (minimum–maximum) for non-normally distributed data. Comparisons between groups were performed using the Independent t-test for normally distributed variables and the Mann-Whitney U test for non-normally distributed variables. Categorical variables were presented as n (%) and compared using Fisher's exact test or the Chi-square test, as appropriate. A p value of <0.05 was considered statistically significant.

The primary outcome of this study was the difference in total fecal SCFA levels between children with and without HD. Secondary outcomes included differences in the individual SCFA components, namely acetate, propionate, butyrate, and valerate.

RESULTS

A total of 17 pediatric subjects were included in this study, consisting of 9 children with HD and 8 children without HD. The baseline characteristics of the study subjects are summarized in **Table 1**.

Table 1. Baseline characteristic of the study subjects

Variables	HD (n=9)	Without HD (n=8)	p-value
Age, median (min-max), months	28 (7-168)	24 (6-71)	0.423
Gender			
Male (%)	7 (77.8)	2 (25)	0.057
Female (%)	2 (22.2)	6 (75)	
Exclusive breastfeeding			
Yes (%)	8 (88.9)	8 (100)	1.000
No (%)	1 (10.1)	0 (0)	
Definitive surgical treatment			
Yes (%)	4 (44.4)	0 (0)	0.082
No (%)	5 (55.6)	8 (100)	
HAEC history			
Yes (%)	4 (44.4)	0 (0)	0.082
No (%)	5 (55.6)	8 (100)	
Nutritional status			
Normal (%)	5 (55.6)	8 (100)	0.098
Undernourished (%)	2 (22.2)	0 (0)	
Severely malnourished (%)	2 (22.2)	0 (0)	

HD: Hirschsprung's disease, Min: Minimum, Max: Maximum, HAEC: Hirschsprung-associated enterocolitis

The characteristics analyzed included age, gender, nutritional status, and relevant clinical variables. There were no significant differences between the HD group and the non-HD group in baseline demographic characteristics (p>0.05), indicating that the two groups were comparable at baseline.

The results of fecal SCFA analysis are summarized in **Table 2**. The comparison between the HD group and the non-HD group showed a significant difference in total SCFA levels.

Table 2. SCFA analysis result data

Variables	HD (n=9)	Without HD (n=8)	p value
Total SCFA levels (mg/ml)	2.4±1.6	7.9±2.4	<0.05[a]
Absolute butyrate levels (mg/ml)	0.9±0.3	0.8±0.4	0.921[b]
Butyrate levels (%)	3 (1-25)	8.5 (3-14)	0.481[b]
Propionate levels (%)	7 (1-21)	21.5 (10-24)	0.300[b]
Valerate levels (%)	0.9 (0.2-2.3)	0.4 (0.1-3.6)	0.309[b]
Acetate levels (%)	78.1±15.3	67.5±7.4	0.095[b]

a: Independent t-test analysis, b: Mann-Whitney analysis. Categorical variables are presented as n (%). Normally distributed numerical variables are presented as mean±standard deviation. Non-normally distributed numerical variables are presented as median (minimum–maximum).
SCFA: Short-chain fatty acid, HD: Hirschsprung's disease

The mean total SCFA level in children with HD was 2.4±1.6. This was significantly lower than that children without HD (7.9±2.4) (p<0.05) (**Table 2**). Further analysis of individual SCFA components showed no significant differences between the two groups. There were no significant differences in absolute butyrate levels, butyrate levels, propionate levels, valerate levels, or acetate levels between children with HD (n=9) and children without HD (n=8) (all p>0.05) (**Table 2**).

DISCUSSION

In this study, fecal total SCFA levels in children with HD (2.4±1.6) were significantly lower than in children without HD (7.9±2.4) (p<0.05). This finding is consistent with the study by Liu et al.,[13] who used Mendelian randomization analysis to investigate the association between gut microbiota and HD. The study demonstrated a reduction in *Clostridiaceae* and *Ruminococcus*, which are protective intestinal flora and major producers of SCFAs, thereby predisposing patients with HD to lower SCFA levels. One factor contributing to alterations in gut microbiota diversity is surgical intervention. Previous studies have shown a reduction in gut microbiota diversity in patients with HD after definitive surgery compared with healthy children, with significant differences in the degree of microbial diversity loss depending on the length of colonic resection.[6]

Reduced SCFA levels in children with HD are also associated with a history of HAEC. Demehri et al.[3] reported that fecal SCFA levels were reduced by more than fourfold in children with a history of HAEC, accompanied by changes in SCFA composition, suggesting a complex interaction between colonic metabolism and alterations in gut microbiota. In HAEC, bacterial overgrowth and reduced microbial diversity increased release of inflammatory factors, resulting in intestinal dysmotility which further promotes bacterial overgrowth and perpetuates microbial imbalance. These processes are mediated by intestinal epithelial changes, including reduced production of secretory phospholipase A2 (sPLA2), increased bacterial lipopolysaccharide (LPS) production, and downregulation of TFF3, SPDEF, and KLF4 expression. This would cause a reduction in goblet cells and decreased neutral and acidic mucin secretion therefore weakening the mucosal barrier function.[14]

In the present study, no significant difference was found in absolute butyrate levels between the HD group (0.9±0.3 mg/ml) and the non-HD group (0.8±0.4 mg/ml) (p>0.05). However, the proportion of butyrate tended to be lower in the HD group (3 [1-25]) compared with the non-HD group (8.5 [3-14]). A similar finding was reported by Liu et al.,[13] who observed a reduction in butyrate levels in children with HD, accompanied by a decrease in *Clostridiaceae* abundance. Plekhova et al.[16] further explained that reduced butyrate production in patients with HD may be influenced by increased lysine catabolism, in which lysine serves as an alternative substrate to carbohydrates (the primary substrate for SCFA production). In addition, reduced levels of N-acetylglucosamine and N-acetylneuraminic acid were associated with impaired carbohydrate digestion due to gut microbiota dysbiosis. These findings are in line with previous studies suggesting that characteristic gut microbiota and SCFA profiles in children with HD may contribute to the development of HAEC.[16]

In patients with HD complicated by HAEC, additional metabolic alterations have been reported. This included increased tyrosine catabolism, reduced degradation of trans-4-hydroxy-L-proline (Hyp), and changes in specific volatile compounds. Hyp is a common product of anaerobic proline transformation utilized by gut microbiota, particularly *Clostridium* spp., which are major butyrate producers. Increased Hyp concentrations may indicate a reduction in Hyp-utilizing microbiota and may partly explain the lower SCFA concentrations observed in patients with HAEC.[15] However, these findings differ from those reported by Prato et al.,[17] who performed metagenomic analysis of fecal samples from children with HD with and without HAEC and found increased butyrate production in children with HAEC.

In contrast to other SCFAs, this study found a tendency toward a higher proportion of valerate in the HD group (0.9 [0.2-2.3]) compared with the non-HD group (0.4 [0.1-3.6]). A similar trend was reported by Demehri et al.,[3] who observed increased valerate and isovalerate levels in HD. In addition, the proportion of acetate in this study tended to be higher in children with HD (78.1±15.3) than in children without HD (67.5±7.4). Different results were reported by Chantakhow et al.,[6] who found reduced acetate levels in children with HD compared with butyrate levels. In cases of HAEC, Demehri et al.[3] reported a significant reduction in acetate levels in children with HAEC compared with those with HD without HAEC. Similar findings were also reported by Prato et al.,[17] who demonstrated reduced acetate production due to impaired development of acetate-producing bacterial colonies in children with HAEC.

Limitations

This study has several important limitations. First, the sample size was small, which may limit statistical power and generalizability; therefore, the findings should be interpreted as exploratory. Second, the study included a wide age range, which may have introduced variability in SCFA profiles because gut microbiota composition changes with age. Third, the HD group was heterogeneous, including both pre- and post-operative patients as well as patients with a history of HAEC, all of which may have influenced SCFA levels. Finally, potential confounding factors such as diet, breastfeeding status, and intestinal transit time were not fully controlled.

CONCLUSION

This study demonstrated that children with HD have significantly lower total fecal SCFA levels compared with children without HD. However, no statistically significant differences were observed in individual SCFA components. These findings suggest that alterations in overall SCFA production may be associated with HD, although further studies with larger sample sizes are needed to confirm these results.

ETHICAL DECLARATIONS

Ethics Committee Approval

This study was conducted with approval from the Ethics Committee of Faculty of Medicine, Universitas Indonesia (Date: 18.12.2023, Decision No: KET-1814/UN2.F1/ETIK/PPM.00.02/2023).

Informed Consent

Informed consent was obtained from a parent or legal guardian. Where appropriate, age-adjusted assent was also obtained from the child. The inclusion of vulnerable populations in this study adhered to national and international ethical guidelines. Extra care was taken to ensure voluntary participation, understanding, and protection of participant dignity and autonomy.

Peer Review Process

This manuscript was subject to external peer review.

Conflict of Interest

The authors declare no conflicts of interest related to this study.

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Author Contributions

Concept: RPT, TAK, YY; Design: RPT, TAK, YY; Control: RPT, TAK, YY; Data Collection and/or Processing: RPT, TAK, YY; Analysis and/or Interpretation: RPT, TAK, YY; Literature Review: RPT, TAK, YY; Article Writing: RPT, TAK, YY; Critical Review: All Authors.

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REFERENCES

1. Kessmann J. Hirschsprung's disease: diagnosis and management. *Am Fam Physician*. 2006;74(8):1319-1322.
2. Bradnock TJ, Knight M, Kenny S, Nair M, Walker GM. Hirschsprung's disease in the UK and Ireland: incidence and anomalies. *Arch Dis Child*. 2017;102(8):722-727. doi:10.1136/archdischild-2016-311872
3. Demehri FR, Halaweish IF, Coran AG, Teitelbaum DH. Hirschsprung-associated enterocolitis: pathogenesis, treatment and prevention. *Pediatr Surg Int*. 2013;29(9):873-881. doi:10.1007/s00383-013-3353-1
4. Frykman PK, Short SS. Hirschsprung-associated enterocolitis: prevention and therapy. *Semin Pediatr Surg*. 2012;21(4):328-335. doi:10.1053/j.sempedsurg.2012.07.007
5. Sakurai T, Tanaka H, Endo N. Predictive factors for the development of postoperative Hirschsprung-associated enterocolitis in children operated during infancy. *Pediatr Surg Int*. 2021;37(2):275-280. doi:10.1007/s00383-020-04784-z
6. Chantakhow S, Khorana J, Tepmalai K, Boonchooduang N, Chattipakorn N, Chattipakorn SC. Alterations of gut bacteria in Hirschsprung disease and Hirschsprung-associated enterocolitis. *Microorganisms*. 2021;9(11):2241. doi:10.3390/microorganisms9112241
7. Morrison DJ, Preston T. Formation of short chain fatty acids by the gut microbiota and their impact on human metabolism. *Gut Microbes*. 2016;7(3):189-200. doi:10.1080/19490976.2015.1134082
8. Cummings JH, Pomare EW, Branch WJ, Naylor CP, Macfarlane GT. Short chain fatty acids in human large intestine, portal, hepatic and venous blood. *Gut*. 1987;28(10):1221-1227. doi:10.1136/gut.28.10.1221
9. Ohira H, Tsutsui W, Fujioka Y. Are short chain fatty acids in gut microbiota defensive players for inflammation and atherosclerosis? *J Atheroscler Thromb*. 2017;24(7):660-672. doi:10.5551/jat.RV17006
10. Rao SC, Esvaran M, Patole SK, et al. Gut microbiota in neonates with congenital gastrointestinal surgical conditions: a prospective study. *Pediatr Res*. 2020;88(6):878-886. doi:10.1038/s41390-020-0824-7
11. Chi C, Xue Y, Lv N, et al. Longitudinal gut bacterial colonization and its influencing factors of low birth weight infants during the first 3 months of life. *Front Microbiol*. 2019;10:1105. doi:10.3389/fmicb.2019.01105
12. Granéli C, Dahlin E, Börjesson A, Arnbjörnsson E, Stenström P. Diagnosis, symptoms, and outcomes of Hirschsprung's disease from the perspective of gender. *Surg Res Pract*. 2017;2017:9274940. doi:10.1155/2017/9274940
13. Liu W, Yan H, Jia W, et al. Association between gut microbiota and Hirschsprung disease: a bidirectional two-sample Mendelian randomization study. *Front Microbiol*. 2024;15:1366181. doi:10.3389/fmicb.2024.1366181
14. Li S, Zhang Y, Li K, et al. Update on the pathogenesis of the Hirschsprung-associated enterocolitis. *Int J Mol Sci*. 2023;24(5):4602. doi:10.3390/ijms24054602
15. Tang W, Su Y, Yuan C, et al. Prospective study reveals a microbiome signature that predicts the occurrence of post-operative enterocolitis in Hirschsprung disease patients. *Gut Microbes*. 2020;11(4):842-854. doi:10.1080/19490976.2020.1711685
16. Plekhova V, De Paepe E, Van Renterghem K, Van Winckel M, Hemeryck LY, Vanhaecke L. Disparities in the gut metabolome of post-operative Hirschsprung's disease patients. *Sci Rep*. 2021;11(1):16167. doi:10.1038/s41598-021-95589-0
17. Prato A, Bartow-McKenney C, Hudspeth K, et al. A metagenomics study on Hirschsprung's disease associated enterocolitis: biodiversity and gut microbial homeostasis depend on resection length and patient's clinical history. *Front Pediatr*. 2019;7:326. doi:10.3389/fped.2019.00326

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A review of the management of prenatally detected fetal anomalies: the need for structured evaluation and a multidisciplinary approach

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ABSTRACT

The evaluation and management of prenatally detected fetal anomalies continue to be a challenging problem. The judicious use of screening tests, determination of severity of a particular disorder and the decision-making regarding prenatal and perinatal management pose unique difficulties. This narrative review aims to addresses the critical aspects with regard to prenatally detected fetal anomalies with respect to their diagnosis, evaluation and management. They are presented in the light of clinical experience and observations with regard to clinical management of these disorders. The key information pertaining to the clinical use of prenatal diagnostic modalities, maternal screening and prenatal and perinatal management are summarized here. The wide spectrum of fetal anomalies range from physiological phenomena of uncertain significance to complex, lethal anomalies. The practical issue with the clinical management of a pregnancy complicated by fetal anomaly lies in the lack of universally accepted guidelines in management. The use of invasive fetal diagnosis, fetal therapy and termination of pregnancy for fetal anomalies continue to be matters of medical, social and ethical concern. The management of prenatally detected fetal anomalies should follow a structured and scientific approach to yield optimal results. The prenatal, perinatal and postnatal management can be thus streamlined. Establishment of standard clinical guidelines, multidisciplinary teams and specialized centers will help to optimize the management of fetal anomalies.

Keywords: Prenatal diagnosis, fetal anomaly, maternal screening, fetal therapy, perinatal management

INTRODUCTION

The evaluation and management of prenatally detected fetal anomalies continue to be a challenging problem. The determination of severity of a particular disorder, selection of the appropriate tests for evaluation and institution of the most appropriate management in a particular scenario require consideration of multiple factors. The new and evolving methods of genetic testing, invasive fetal diagnostic techniques and advances in fetal imaging have contributed to an early and more precise diagnosis. But these advancements are also complicated by concerns regarding their interpretation, significance and appropriate, scientific use.

The practical issue with the clinical management of a pregnancy complicated by fetal anomaly lies in the lack of universally accepted guidelines in the diagnosis, evaluation and therapy. The wide spectrum of these disorders range from physiological phenomena of uncertain significance to complex, lethal anomalies, that can not be fitted under a single umbrella. The use of invasive fetal diagnosis, fetal therapy and termination of pregnancy for fetal anomalies (TOPFA) continue to be matters of medical, social and ethical concern. Moreover, the best possible outcome ideally requires the input of multiple specialists, a scientific and structured approach and multidisciplinary teams. These facts point to the need to establish universal standard treatment guidelines and clinical practice principles. There is also a need for institution of medical boards and specialized centers to streamline the management of fetal anomalies.

Aims

This article aims to review the various aspects of the diagnosis, evaluation and management of prenatally detected fetal anomalies. The essential objective is to provide a comprehensive overview of prenatal screening, fetal therapy,

prenatal and perinatal management. The article also aims to propose future directions towards standardization of the practice of fetal medicine.

Methods

A narrative review of the present literature, clinical guidelines and practice recommendations is performed here in the light of clinical experience and observations with regard to clinical management of these variegated group of disorders. Current literature including PubMed, Scopus, Embase and Medline database articles, clinical guidelines and practice recommendations from 2004-2024 were reviewed. The focus mainly rested on articles pertaining to the 'prenatal diagnosis', 'TOPFA', 'fetal therapy' and evaluation and management of 'fetal anomalies'. An attempt has been made to synthesize and organize the said data about various aspects of fetal anomalies for easy reference. The key observations pertaining to the clinical application of these principles are also summarized here.

Main Text

When faced with the issue of treatment of fetal anomalies, there are numerous concerns to be addressed by the concerned specialists. These pertinent factors with regard to diagnosis, evaluation and management of fetal anomalies are summarized in the **Table 1**. The various aspects of this subject is discussed here under the following headings:

- The optimal use of presently available maternal screening tests
- The use of invasive fetal diagnosis, fetal therapy and EXIT procedure
- The decision-making and communication with regard to fetal anomalies
- The perinatal management with respect to fetal anomalies
- Fetal anomalies and medical termination of pregnancy (MTP)
- The need for a multidisciplinary and integrated approach to management
- The need for specialized organizational entities for fetal anomalies

THE OPTIMAL USE OF PRESENTLY AVAILABLE MATERNAL SCREENING TESTS

First trimester screening is performed with maternal serum biomarkers, pregnancy-associated plasma protein A (PAPP-A) and free beta-human chorionic gonadotropin (hCG). Second trimester screening is performed with alfa fetoprotein (AFP), hCG, estriol, and inhibin-A, the "quad screen". These are useful in the detection of chromosomal disorders, neural tube defects (NTD) etc. Carrier genetic screening is also available to detect inherited disorders for high-risk cases (pre-implantation genetic testing).[1-3]

Cell free fetal DNA study by isolation of fetal cells in the maternal circulation using advanced sorting techniques permit genetic testing from a maternal blood sample. It can be done from 10 weeks of pregnancy and beyond. This can be used for the screening of trisomy 21, 18, 13 and sex-chromosome aneuploidy. This is only a screening tool and a positive cell-free DNA test result should be followed by a diagnostic test with amniocentesis or chorionic villus sampling (CVS). The high cost factor, technical complexity, the lack of standardization and non-uniform insurance cover for tesing limit the widespread adoption of this test.[4-7]

Prenatal genetic testing: Prenatal genetic testing techniques to detect aneuploidy include QF-PCR and FISH for rapid aneuploidy testing, G-banded karyotyping and chromosomal microarray analysis (CMA). The adoption of next generation sequencing (NGS) techniques in prenatal diagnostics, including whole exome sequencing (WES) and whole genome sequencing (WGS) enable a much greater diagnostic yield.[8-11]

The prenatal diagnosis of a lethal genetic disorder enables appropriate prenatal, perinatal and postnatal management, and also termination of pregnancy where indicated. A proper genetic diagnosis also permits counselling about the risk of recurrence and facilitates prenatal testing or pre-implantation genetic diagnosis in subsequent pregnancies.[9-11]

Prenatal genetic screening (serum screening with or without nuchal translucency (NT) US/ cell-free DNA screening) and diagnostic testing (CVS or amniocentesis) options should be discussed with all parents during pregnancy, regardless of maternal age or risk of chromosomal abnormality. The

Table 1. The 'six pertinent questions each' with regard to the prenatal diagnosis, evaluation and therapy of a pregnancy complicated by fetal anomaly

Prenatal diagnosis	Prenatal evaluation	Prenatal and perinatal therapy
The need for carrier screening and use of maternal screening tests	The reliability of the diagnosis and the need for confirmatory testing	Indication for non-invasive/ medical therapy (steroids for fetal lung maturation)
The nature of anomaly: developmental aberration/ significant defect	The need for only routine scans/use of serial testing	Indication for invasive fetal therapy: endoscopic/ surgical
Severity of the anomaly and grading of the defect	The need for detailed fetal evaluation (fetal echocardiogram, fetal MRI etc.)	Indication for EXIT procedure
Isolated defect or part of a complex association	The need for invasive prenatal diagnosis (amniocentesis, CVS etc.)	Indication for termination of pregnancy for fetal anomaly
Likely natural history, progression and prognosis	The need for invasive fetal testing (fetal cord blood or urine sampling etc.)	Indication for referral to a dedicated perinatal centre
Likely complications: prenatal, perinatal and postnatal/fetal (hydrops fetalis, IUGR etc.), maternal (PIH) and gestational (oligohydramnios)	Maternal evaluation for specific risk factors like pre-eclampsia.	Indication for change in perinatal therapy: preterm delivery/caesarean section

MRI: Magnetic resonance imaging, CVS: Chorionic villus sampling

parents retain the right to pursue or decline prenatal genetic screening and diagnostic testing, after proper counselling. If screening is accepted, patients should have one prenatal screening approach, and should not have multiple screening tests performed simultaneously. The sensitivity of first trimester and second trimester screening tests are 80-84% and 80-82% respectively. The specificity of first trimester and second trimester screening tests are both about 95%.[9,11-13]

First trimester ultrasound scan (US) at 11 to 13 weeks is very useful as it aids in precise dating, detection of twin chorionicity, and the early detection of major structural abnormalities. Nuchal translucency (NT) measurement on US at 11 to 14 weeks is a marker for fetal chromosomal disorders, NTD, structural anomalies (cardiac) and genetic disorders.[14-16]

Second trimester US is vital to assess major defects of the heart, brain and spine, kidneys, abdomen, craniofacial region, limbs etc. This is to be done at 18 to 22 weeks of pregnancy. Detailed fetal imaging using 4D US, fetal echocardiogram, fetal MRI and serial US are indicated in complex anomalies.[14-17]

Parents with a positive screening test result for fetal aneuploidy should undergo genetic counselling and a comprehensive US evaluation with an opportunity for diagnostic testing (CVS, amniocentesis) to confirm results. Parents with a negative screening test result should be informed that the result indicates significantly lower risk of the specific aneuploidy. But this does not fully ascertain that the fetus is unaffected. Fetal structural defects may occur with or without fetal aneuploidy. Hence all patients should be offered a second-trimester US between 18 and 22 weeks of gestation (with or without second-trimester maternal serum AFP).[1,8,11,15,17]

THE USE OF INVASIVE FETAL DIAGNOSIS, FETAL THERAPY AND EXIT PROCEDURE

Invasive fetal diagnosis relies on CVS in the first trimester and amniocentesis in the second trimester. These are selectively used in the case of a positive screening test and also in high-risk cases, for the sake of a definitive diagnosis. These are complicated by the attendant risks to the mother and fetus. CVS is mainly used for diagnosis of genetic disorders. Amniocentesis aids in the evaluation of chromosomal, biochemical, histopathological, and infective disorders.[18-21] The common prenatal investigations, their ideal timing and their general significance are summarized in the **Table 2.**

Fetal therapy: Most of the advanced fetal therapy is still experimental and generally conducted only as a part of clinical trials, in selected high-risk cases. The benefits of intervention should outweigh the risks posed to the mother and fetus. Fetal endoscopy (FETENDO), fetal transfusion and open surgery are used in select cases. The maternal administration of drugs like steroids benefit the fetus in certain situations like lung disorders.[2,16,18,21]

EXIT procedure is reserved for fetal anomalies that benefit from intrapartum intervention. These include conditions like congenital high airway obstruction syndrome (CHAOS) that mandate early airway access and conditions requiring immediate cannulation for ECMO. The EXIT procedure is associated with a higher maternal risk than a caesarean. There are also considerable logistical challenges due to the need for multidisciplinary involvement. The decision regarding the use of invasive fetal diagnosis, fetal therapy and EXIT procedure need careful risk-benefit assessment. This decision should be taken by a team involving all the concerned specialists.[2,16,19,21]

THE DECISION-MAKING AND COMMUNICATION WITH REGARD TO FETAL ANOMALIES

The salient points in the maternal history with relevance to fetal anomaly include the following points: a. history of consanguinity, b. affected siblings, c. previous fetal loss, d. advanced maternal age, e. treatment for infertility, f. bad obstetric history, g. maternal illness/carrier state, h. history of genetic disorder and i. risk of recurrence of anomaly in the subsequent pregnancies. These should be surveyed, analysed and documented. In case of detection of multiple anomalies,

Table 2. Summary of the common prenatal investigations, their timing and general significance

Prenatal testing	The investigation	Significance
Carrier testing	Genetic testing done before pregnancy.	Detection of inherited disorders in high risk cases
Prenatal genetic testing to detect aneuploidy	QF-PCR and FISH, G-banded karyotyping CMA, NGS: WES and WGS	Diagnosis of genetic defects
First trimester screening	Maternal serum biomarkers, PAPP-A and free beta hCG	Detection of chromosomal disorders, neural tube defects, nuchal translucency measurement on US etc.
Cell free fetal DNA testing	Genetic testing from maternal blood sample from 10 weeks and beyond	Used for the screening of trisomy 21, 18, 13 and sex-chromosome aneuploidy.
First trimester scan	Ultrasound at 11-13 weeks	Precise dating, detection of twin chorionicity, and early detection of major structural anomalies.
Second trimester screening	Quad screen with AFP, hCG, estriol, and inhibin-A	Detection of chromosomal disorders, neural tube defects etc.
Second trimester scan	Ultrasound at 18-22 weeks	Assess major defects of the heart, brain and spine, kidneys, abdomen, craniofacial region, limbs etc.
Fetal diagnostic testing	Chorionic villus sampling in first trimester and amni-ocentesis in the second trimester	Selectively used for a positive screening test and high-risk cases for definitive diagnosis
Fetal sampling	Fetal cord blood sampling fetal urine sampling	Diagnosis of TORCH infections, prognostication PUV etc.

PAPP-A: Pregnancy associated plasma protein A, hCG: Human chorionic gonadotropin, QF-PCR: Quantitative fluorescence polymerase chain reaction, FISH: Fluorescence in situ hybridization, CMA: Chromosomal microarray, NGS: Next generation sequencing, WES: Whole exome sequencing, WGS: Whole genome sequencing, PUV: Posterior urethral valves

the management decisions should be based on the major issue with the most significant bearing on the outcome and issues that affect perinatal therapy (major cardiac defects).[2,16,19-21]

The parents should receive education, counselling and psychological support during the prenatal and postnatal period. The communication should be compassionate, empathetic and open. The objective is to help them understand the situation and arrive at an informed decision. The important factors with regard to this communication pertain to the following points: a. nature of the anomaly and it's severity, b. the likely prognosis and outcome, c. the need for further investigations, d. the option of TOPFA, where indicated, e. the need for invasive diagnostic/therapeutic modalities and, f. the need for change in mode, time or venue of delivery.[2,16,19-21]

The obstetrician is usually the first contact of parents in case of prenatally detected anomalies. They should refer the mother to the appropriate center for the fetal specialist to arrive at a diagnosis, assess the prognosis and decide on the need for further evaluation. The initial communication regarding the possible nature of the anomaly and management should ideally be made by the fetal specialist. The sonologist/obstetrician may not have received specific training in fetal medicine to provide a definite diagnosis and prognosis. The management plan should be formulated by the team of specialists. A continuous and dynamic interaction should be maintained between the obstetrician and the fetal specialist during the follow up and serial re-evaluation. This enables to update decisions regarding antenatal and perinatal management, keeping the parents abreast with the progress.[12,13,16,19-21]

THE PERINATAL MANAGEMENT WITH RESPECT TO FETAL ANOMALIES

After the diagnosis of a fetal anomaly, the appropriate perinatal management may require transfer to a specialised centre/involvement of multiple specialists. Though broad guidelines can be followed in the management, decision-making in each case of fetal anomaly has to be individualised. The ideal timing and mode of delivery are also to be decided upon based on risk-benefit assessment. Transporting a pregnant woman before delivery to a hospital that can provide the necessary level of care is a much safer option than transferring an unstable neonate who requires urgent intervention in the immediate postnatal period.[22-25]

Anomalies in the fetus can affect gestation and vice versa, highlighting the vital need for close interaction and continued communication between the obstetrician and the fetal specialist during the follow up. Lack of such coordination and planned decision-making exposes both the mother and the child to avoidable stress and complications.[11-15] The timing and mode of delivery are important considerations in all major fetal anomalies. But these are especially vital in conditions characterized by a complicated perinatal period like cardiac defects, congenital diaphragmatic hernia (CDH), oesophageal atresia, abdominal wall defects, NTD etc. Spontaneous vaginal delivery should be the primary option in most fetal anomalies. This is associated with lowest maternal morbidity and mortality.[24-27]

In case of lethal anomalies that have not undergone TOPFA, and have reached late gestation, term vaginal delivery is to be preferred, as this is the safest option for the mother. Caesarean delivery is generally indicated if a fetal anomaly is associated with the risk of dystocia, bleeding, or disruption of a protective sac. Examples of the same include anomalies like large exomophalos, severe hydrocephalus, large myelomeningocele (MMC), and teratomas.[22,24-26]

In many situations with prenatal diagnosis of fetal anomalies, there is a tendency for Obstetricians to proceed with delivery immediately after completion of 36 weeks. But it is ideal to wait for delivery till 38-39 weeks to optimise the risk factors, unless gestational factors supervene. It should be remembered that the fetus in utero is at lesser risk and better protected than a preterm neonate in the intensive care unit. The fact that mortality and morbidity in preterm neonates with anomalies is higher than term neonates is worth consideration here. Due to the severe risks related to it, preterm delivery should be reserved for few selected cases of fetal anomalies, the most common indication being the worsening of fetal status.[22,23,25,27]

Planned labour is of utmost importance in a neonate with a major structural anomaly (CDH, oesophageal atresia, cardiac defects, abdominal wall defects etc.). If an unplanned delivery occurs outside regular hours in these conditions, the well-being of the neonate is critically endangered. This is primarily due to logistical constraints pertaining to the availability of neonatologists and paediatric surgeons. Planned induction of labour is also beneficial for mothers who reside far away from a tertiary care centre. A broad outline of the evaluation and management pathways in pregnancy complicated by fetal anomalies is summarised in the **Figure**.[23,26-28]

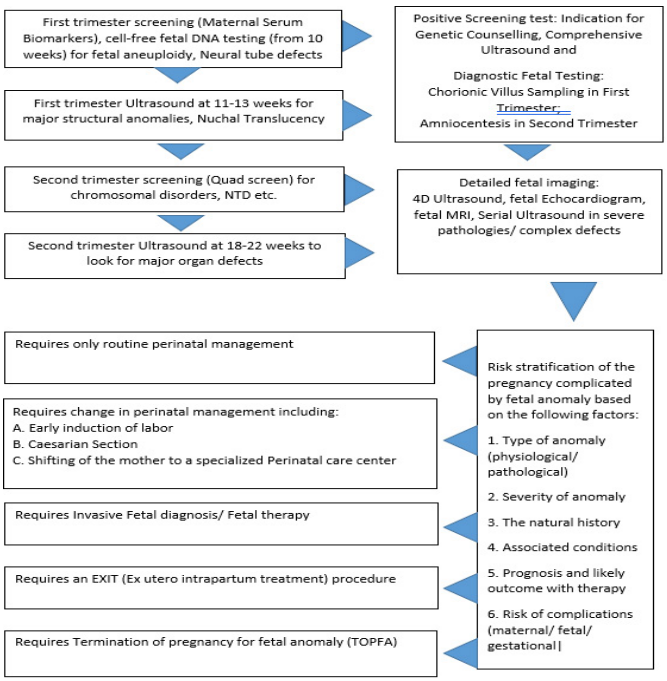

Figure. A broad and general outline of the pathway for evaluation and management in pregnancy complicated by fetal anomalies

FETAL ANOMALIES AND MEDICAL TERMINATION OF PREGNANCY (MTP)

Widespread use and advancements in prenatal diagnostic techniques have resulted in increased detection of anomalies, with an increase in demand for TOPFA. In case of major anomalies that are detected in the first trimester (anencephaly, pulmonary agenesis, body stalk anomaly etc.), TOPFA should be considered at the earliest. The relaxation in the legal gestational age limits for TOPFA in various countries has eliminated an important barrier in this regard. There have also been directions for a medical board to be established for special permission with regard to MTP in specific situations. In many countries, the gestational upper limit has been increased to 24 weeks and beyond 24 weeks, with the permission of the medical board, with no gestational upper limit in the cases of major fetal congenital malformations. Fetal anomalies may sometimes be recognised only after 20 weeks of gestation and continuation of such a pregnancy could be detrimental to the mother and the foetus. In such situations, the woman can approach the medical board for an MTP.[2,16,29-32]

Delay in diagnosis of a fetal anomaly due to factors like delay in seeking antenatal care, delayed diagnosis, irregular follow-up, inadequate counselling and late referral are the major reasons for the delay in the decision for abortion. Abortion-related complications increase with gestational age. Also, the pregnancy termination late in gestation has the inherent risk for the occurrence of a live birth. This can result in great anguish for parents and also become a severe drain on healthcare resources.[30-32] Hence, in late gestation, a feticide procedure may be required for prevention of occurrence of a live birth. Feticide must be considered before MTP for gestation that has progressed beyond 24 weeks. It is the responsibility of the health care sector to facilitate access to early and effective prenatal diagnosis. This would help to reduce the number of cases where TOPFA needs to be considered late in the gestation.[19-21,29-32]

THE NEED FOR A MULTIDISCIPLINARY AND INTEGRATED APPROACH TO MANAGEMENT

The essential objective in the case of prenatal detection of a fetal anomaly is to provide the pregnant woman with comprehensive specialized care. A fetal specialist (maternal-fetal medicine specialist/paediatric surgeon) confirms the anomaly and determines the prognosis and provides therapeutic options. The opinion regarding severity and prognosis of a fetal anomaly can be organ-specific and may need the input of multiple specialists.[33-36]

The paediatric and neonatal surgeon is in a pivotal position to opine regarding the diagnosis, prognosis and therapy with regard to most of the surgically treated structural anomalies. The opinion of other specialists should be integrated in the decision-making process. The use of technology for image sharing and collaboration can help to overcome the logistical challenges of garnering multiple opinions. It is imperative that the fetal specialist/paediatric surgeon be abreast with all the new diagnostic/therapeutic techniques, clinical guidelines and the current medico legal developments in this regard.[2,16,32-36]

The input of other specialists like cardiology, neurology, nephrology, genetics, orthopedics and paediatrics is vital in certain specific disorders involving specific organ systems. The team of specialists should decide on the evaluation, frequency of follow-up, the mode, timing and venue of the delivery. The obstetrician, in consultation with the paediatric surgeon and neonatologist, suggests the ideal prenatal, perinatal and postnatal management in each case.[33-36]

THE NEED FOR SPECIALIZED ORGANIZATIONAL ENTITIES

The factors which are narrated in this article reiterate the need for establishment of two separate organizational entities: 1. Specialized centers for facilities for antenatal diagnosis, evaluation and follow up, with multidisciplinary teams: Advanced facilities for fetal diagnosis should be available at these centers. The involvement of concerned specialists, whenever necessary, will help to improve diagnostic accuracy and streamline management. The paediatric surgeon/fetal medicine specialist could act as the central person coordinating evaluation, communications and decisions. This group can constitute the medical board that has to be involved in contentious decision making in pregnancy complicated by fetal anomaly. The prenatal follow up by the surgeon who is responsible for postnatal care can ease communication, facilitate decision-making and improve confidence of the parents.[2,16,33-36]

2. Specialized perinatal centers with the advanced facilities for EXIT, neonatal medicine and neonatal surgery: Mothers with fetus having major anomalies with high risk of complications need specialist medical/surgical therapy during perinatal period. They can be referred at the appropriate time during gestation to such centers for advanced care. Delivery can also be conducted at hospitals in close proximity to these specialized centers, so that transport-related issues can be minimized.[2,16,19-21,33-36]

DISCUSSION

The rapid advancements and widespread use of prenatal genetic testing and fetal imaging have greatly enhanced the diagnostic yield of fetal anomalies. Newer NGS techniques like WES have greatly improved diagnostic accuracy. But these have also resulted in detection of anomalies of uncertain clinical significance. This should be an important consideration while dealing with all fetal anomalies. The decisions regarding the use of screening studies (aneuploidy screening) should be taken considering factors like maternal risk, parental request, medical indications and local regulations, in consultation with concerned specialists whenever necessary. The role of a pediatrician and geneticist is crucial in this regard. It is also vital that all the concerned specialists involved in the management of fetal anomalies be well versed with the latest guidelines and diagnostic options.

The advances in fetal therapy have not generally kept pace with the rapid strides in fetal diagnosis, thus limiting the

options of prenatal therapy. These facilities may be available in only select centres, for high-risk cases, as a part of clinical trials. But the advancements in therapeutic options and fetoscopic techniques may lead to increased adoption of fetal therapy in the future. The present scenario dictates the need for institution of the ideal perinatal therapy for each case, especially with regard to the venue, timing and mode of delivery. The issues pertaining to MTP, TOPFA, invasive fetal diagnosis and fetal therapy have medico legal, ethical and social ramifications, in addition to the medical considerations. Moreover, the issue of MTP is associated with cultural concerns and legal framework-related implications which can be locally variable. These facts reiterate the need for the involvement of a multidisciplinary team in taking management decisions.

The optimal use of fetal diagnostic and therapeutic modalities and scientific perinatal management depend on the accurate estimation of severity of the disorder and the likely complications. The inherent issues with regard to the management of fetal anomalies can only be addressed by the formulation of standard guidelines and adoption of a multidisciplinary approach. The establishment of global registries and collaborative network of fetal specialists will help to set universally accepted standards of care.

CONCLUSION

The prenatal diagnosis of a fetal anomaly should prompt the involvement of a multidisciplinary team to decide on a comprehensive treatment plan. The paediatric surgeons have a pivotal role to play in this regard in consideration of their expertise in the treatment of congenital disorders. The primary objective of the collaborative approach is to ensure structured evaluation and timely prenatal, perinatal and postnatal management. The establishment of specialized prenatal and perinatal centers with diagnostic and therapeutic facilitates and trained specialists can go a long way towards achieving this objective.

ETHICAL DECLARATIONS

Peer Review Process

This review was externally peer-reviewed.

Conflict of Interest

The authors declare no conflicts of interest.

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REFERENCES

1. Carlson LM, Vora NL. Prenatal diagnosis: screening and diagnostic tools. *Obstet Gynecol Clin North Am*. 2017;44(2):245-256. doi:10.1016/j.ogc.2017.02.004
2. WHO. Congenital Anomalies [Internet]. Who.int. World Health Organization: WHO; 2016. Available from: https://www.who.int/news-room/fact-sheets/detail/congenital-anomalies
3. Practice Bulletin No. 163: Screening for fetal aneuploidy. *Obstet Gynecol*. 2016;127(5):e123-e137. doi:10.1097/AOG.0000000000001406
4. Levy B, Stosic M. Traditional prenatal diagnosis: past to present. *Methods Mol Biol*. 2019;1885:3-22. doi:10.1007/978-1-4939-8889-1_1
5. Bianchi DW, Chiu RWK. Sequencing of circulating cell-free DNA during pregnancy. *N Engl J Med*. 2018;379(5):464-473. doi:10.1056/NEJMra1705345
6. Qin Y, Yao Y, Liu N, et al. Prenatal whole-exome sequencing for fetal structural anomalies: a retrospective analysis of 145 Chinese cases. *BMC Med Genomics*. 2023;16(1):262. doi:10.1186/s12920-023-01697-3
7. Wang Y, Yin F, Chai Y, et al. Prenatal diagnosis of fetuses with ultrasound anomalies by whole-exome sequencing in Luoyang city, China. *Front Genet*. 2024;14:1301439. doi:10.3389/fgene.2023.1301439
8. Abel DE, Alagh A. Benefits and limitations of noninvasive prenatal aneuploidy screening. *JAAPA*. 2020;33(4):49-53. doi:10.1097/01.JAA.0000654208.03441.23
9. Suciu I, Galeva S, Abdel Azim S, Pop L, Toader O. First-trimester screening-biomarkers and cell-free DNA. *J Matern Fetal Neonatal Med*. 2021;34(23):3983-3989. doi:10.1080/14767058.2019.1698031
10. UK National Screening Committee. Fetal anomaly screening programme–screening for Down's syndrome: UK NSC policy recommendations 2007–2010: model of best practice. 2008. (Department of Health Policy Guideline No. DH_084731).
11. Pan M, Huang LY, Zhen L, Li DZ. A cost-effectiveness analysis comparing two different strategies in advanced maternal age: combined first-trimester screening and maternal blood cell-free DNA testing. *Taiwan J Obstet Gynecol*. 2018;57(4):536-540. doi:10.1016/j.tjog.2018.06.011.
12. Phadke SR, Puri RD, Ranganath P. Prenatal screening for genetic disorders: suggested guidelines for the Indian Scenario. *Indian J Med Res*. 2017;146(6):689-699. doi:10.4103/ijmr.IJMR_1788_15
13. Arslan E, Büyükkurt S, Sucu M, et al. Detection of major anomalies during the first and early second trimester: single-center results of six years. *J Turk Ger Gynecol Assoc*. 2018;19(3):142-145. doi:10.4274/jtgga.2017.0125
14. Paladini D, Volpe P. Ultrasound of congenital fetal anomalies: differential diagnosis and prognostic indicators. 2nd ed. Boca Raton, FL: CRC Press, Taylor & Francis Group; 2014.
15. Pathak S, Lees C. Ultrasound structural fetal anomaly screening: an update. *Arch Dis Child Fetal Neonatal Ed*. 2009;94(5):F384-390. doi:10.1136/adc.2008.137919
16. Buijtendijk MF, Bet BB, Leeflang MM, et al. Diagnostic accuracy of ultrasound screening for fetal structural abnormalities during the first and second trimester of pregnancy in low-risk and unselected populations. *Cochrane Database Syst Rev*. 2024;5(5):CD014715. doi:10.1002/14651858.CD014715.pub2
17. Zaputović SB, Stanojevic M, Honemeyer U, Turudić TP, Kurjak A, Chervenak F. Surgically correctable fetal anomalies: ultrasound diagnosis and management. *Donald School J Ultrasound Obstet Gynecol*. 2012;6(3):237-256. doi:10.5005/jp-journals-10009-1248
18. Coleman BG, Adzick NS, Crombleholme TM, et al. Fetal therapy: state of the art. *J Ultrasound Med*. 2002;21(11):1257-1288. doi:10.7863/jum.2002.21.11.1257
19. Corsello G, Giuffrè M. Congenital malformations. *J Matern Fetal Neonatal Med*. 2012;25(Suppl 1):25-29. doi:10.3109/14767058.2012.664943
20. Bianci D, Crombleholme TM, Dalton ME, Malone FD. Fetology: diagnosis and management of the fetal patient. 2nd ed. New York, NY: McGraw-Hill Medical; 2010.
21. Bhide P, Gund P, Kar A. Prevalence of congenital anomalies in an Indian maternal cohort: healthcare, prevention, and surveillance implications. *PLoS One*. 2016;11(11):e0166408. doi:10.1371/journal.pone.0166408
22. Sadlecki P, Walentowicz-Sadlecka M. Prenatal diagnosis of fetal defects and its implications on the delivery mode. *Open Med (Wars)*. 2023;18(1):20230704. doi:10.1515/med-2023-0704

23. Bijma HH, Schoonderwaldt EM, van der Heide A, Wildschut HI, van der Maas PJ, Wladimiroff JW. Ultrasound diagnosis of fetal anomalies: an analysis of perinatal management of 318 consecutive pregnancies in a multidisciplinary setting. *Prenat Diagn*. 2004;24(11):890-895. doi:10.1002/pd.883
24. Bardi F, Bergman JEH, Siemensma-Mühlenberg N, Elvan-Taşpınar A, de Walle HEK, Bakker MK. Prenatal diagnosis and pregnancy outcome of major structural anomalies detectable in the first trimester: a population-based cohort study in the Netherlands. *Paediatr Perinat Epidemiol*. 2022;36(6):804-814. doi:10.1111/ppe.12914
25. Mangla M, Anne RP. Perinatal management of pregnancies with fetal congenital anomalies: a guide to obstetricians and pediatricians. *Curr Pediatr Rev*. 2024;20(2):150-165. doi:10.2174/1573396318666221005142001
26. Colby CE, Carey WA, Blumenfeld YJ, Hintz SR. Infants with prenatally diagnosed anomalies: special approaches to preparation and resuscitation. *Clin Perinatol*. 2012;39(4):871-887. doi:10.1016/j.clp.2012.09.012
27. ACOG Committee Opinion No. 764: Medically indicated late-preterm and early-term deliveries. *Obstet Gynecol*. 2019;133(2):e151-e155. doi:10.1097/AOG.0000000000003083
28. Craigo SD. Indicated preterm birth for fetal anomalies. *Semin Perinatol*. 2011;35(5):270-276. doi:10.1053/j.semperi.2011.05.011
29. Heaney S, Tomlinson M, Aventin Á. Termination of pregnancy for fetal anomaly: a systematic review of the healthcare experiences and needs of parents. *BMC Pregnancy Childbirth*. 2022;22(1):441. doi:10.1186/s12884-022-04770-4
30. Maurice P, Letourneau A, Benachi A, Jouannic JM. Feticide in second- and third-trimester termination of pregnancy for fetal anomalies: results of a national survey. *Prenat Diagn*. 2019;39(13):1269-1272. doi:10.1002/pd.5594
31. Remez L, Mayall K, Singh S. Global developments in laws on induced abortion: 2008-2019. *Int Perspect Sex Reprod Health*. 2020;46(Suppl 1):53-65. doi:10.1363/46e0920
32. Mohaupt SM, Sharma KK. Forensic implications and medical-legal dilemmas of maternal versus fetal rights. *J Forensic Sci*. 1998;43(5):985-992.
33. Luks FI, Carr SR, Feit LR, Rubin LP. Experience with a multidisciplinary antenatal diagnosis and management model in fetal medicine. *J Matern Fetal Neonatal Med*. 2003;14(5):333-337. doi:10.1080/jmf.14.5.333.337
34. Li G, Yang N, Xie M, et al. Perinatal and follow-up outcome study of fetal anomalies with multidisciplinary consultation. *Ther Clin Risk Manag*. 2017;13:1303-1307. doi:10.2147/TCRM.S138808
35. Kilby MD, Pretlove SJ, Bedford Russell AR. Multidisciplinary palliative care in unborn and newborn babies. *BMJ*. 2011;342:d1808. doi:10.1136/bmj.d1808
36. Raboei EH. The role of the pediatric surgeon in the perinatal multidisciplinary team. *Eur J Pediatr Surg*. 2008;18(5):313-317. doi:10.1055/s-2008-1038641

MHA MEDIHEALTH ACADEMY

Surgery on Children

Case Report

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Delayed diagnosis of distal urethral stricture after hypospadias repair in a child: the critical role of physical examination in preventing irreversible renal damage

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ABSTRACT

We present a pediatric case of distal urethral stricture following hypospadias repair, in which prolonged misattribution of lower urinary tract symptoms to functional causes led to delayed diagnosis and irreversible renal damage. A 12-year-old boy with a history of hypospadias repair in infancy presented with urinary incontinence, incomplete bladder emptying, and nocturia. He had previously undergone multiple pharmacologic treatments without improvement. Physical examination revealed a narrowed coronal meatus. Ultrasonography demonstrated bilateral hydronephrosis and bladder wall thickening, while uroflowmetry showed a plateau-shaped curve with severe obstruction. Intraoperatively, the meatus could only be passed with a 4-Fr tube. Because the meatus was at the coronal level, a dorsal plate incision was extended to the glans tip, providing an adequate flap for tubularized incised plate urethroplasty (TIPU). No graft was required. Postoperatively, uroflowmetry normalized and hydronephrosis regressed, but dimercaptosuccinic acid (DMSA) scintigraphy revealed bilateral renal scars. This case highlights the importance of physical examination and uroflowmetry in the long-term follow-up of children after hypospadias repair, as timely recognition of meatal or urethral obstruction can prevent irreversible upper urinary tract damage.

Keywords: Hypospadias repair, urethroplasty, urethral stricture, hydronephrosis, physical examination

INTRODUCTION

Urinary incontinence in children is most often attributed to functional bladder disorders.[1] However, exclusive focus on functional etiologies without thorough evaluation for anatomical abnormalities may result in delayed diagnosis and irreversible upper urinary tract damage. Distal urethral stricture is a rare but important late complication of hypospadias repair, with reported incidences ranging from 5% to 15%.[2]

The European Association of Urology (EAU) guidelines recommend long-term follow-up after hypospadias repair, including uroflowmetry and meatal calibration even in asymptomatic patients.[3,4] This case illustrates the consequences of overlooking anatomical assessment and underscores the critical role of physical examination in early detection.

CASE

A 12-year-old male refugee, who had fled from a war zone, presented to our department with complaints of daytime and nighttime urinary incontinence, incomplete bladder emptying, and increased nocturnal frequency. He had undergone hypospadias repair in infancy, but the exact date, location, and surgical technique were unknown due to lack of records. There was no history of urinary tract infection.

Over the years, the patient had been treated at different centers for presumed functional bladder dysfunction, receiving various combinations of anticholinergics, desmopressin, and imipramine, without significant symptom improvement.

On presentation, physical examination revealed a narrowed coronal meatus. Renal and bladder ultrasonography demonstrated bilateral hydronephrosis (right AP (anteroposterior) diameter 23 mm, left 19 mm) and bladder wall thickening (20 mm). Preoperative uroflowmetry showed a plateau-shaped curve, Qmax (maximum flow rate) 2.1 ml/s, Qave (average flow rate) 1.4 ml/s, voided volume 356 ml, and post-void residual urine of 260 ml (**Figure 1**).

During surgery, the meatus could only be passed with difficulty using a 4-Fr feeding tube. A dorsal plate incision was performed to enlarge the opening, after which

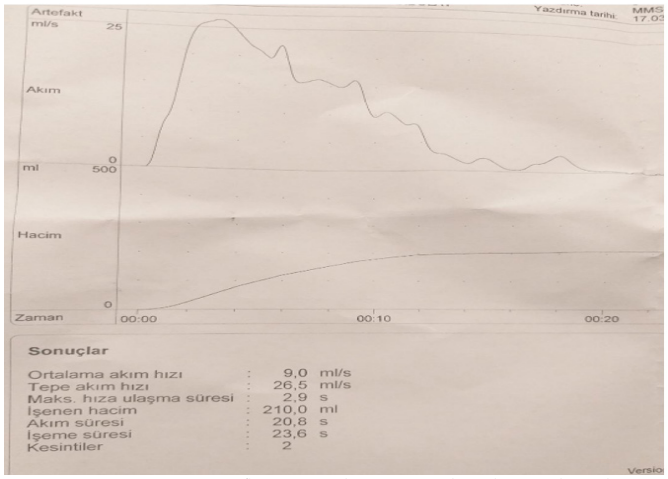

Figure 1. Preoperative uroflowmetry demonstrated a plateau-shaped curve consistent with severe obstruction

cystoscopy was carried out. Because the meatus was located at the coronal level, a dorsal plate incision was extended to the glans tip before cystoscopy, which provided an adequate flap for tubularized incised plate urethroplasty (TIPU). No graft was required.[6,8] The bladder mucosa exhibited minimal trabeculation, both ureteral orifices were in normal location and appearance, no posterior urethral valves were present, and no other strictures were identified. Following cystoscopy, TIPU urethroplasty was completed. A 12-Fr foley bladder catheter was inserted and removed on postoperative day 5; the patient subsequently voided with a strong urinary stream originating from the meatal tip.

At the 1-month follow-up, ultrasonography showed marked regression of hydronephrosis, with complete bladder emptying. Postoperative uroflowmetry revealed a normal bell-shaped curve with Qmax 26.5 ml/s, Qave 9.0 ml/s, voided volume 210 ml, and no residual urine (**Figure 2**). DMSA (dimercaptosuccinic acid) scintigraphy revealed bilateral renal scarring with split renal function of 63% (left) and 37% (right). During the postoperative follow-up, the child was also evaluated by pediatricians at another hospital. As part of their assessment, a voiding cystourethrogram (VCUG) was performed, which showed no signs of vesicoureteral reflux. Unfortunately, long-term follow-up was not possible as the family returned to their home country.

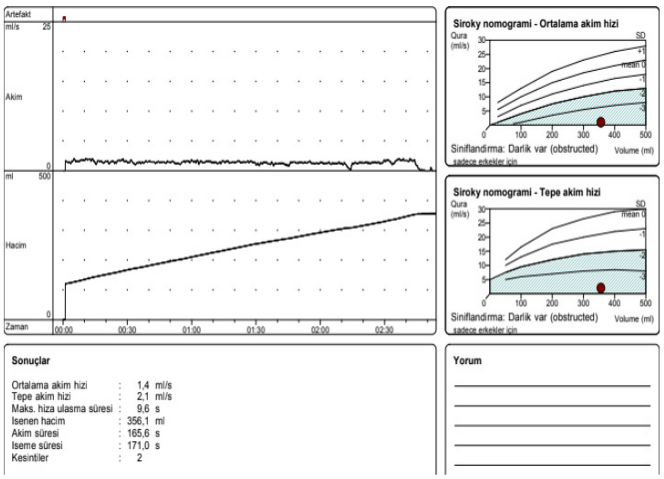

Figure 2. Postoperative uroflowmetry demonstrated a bell-shaped curve indicating normalized urinary flow

DISCUSSION

This case highlights how prolonged misinterpretation of urinary symptoms as purely functional in origin can delay the identification of a surgically correctable anatomical abnormality. The patient had undergone years of medical therapy without benefit, when a simple physical examination could have led to the correct diagnosis earlier.

Distal urethral stricture after hypospadias repair, although uncommon, can cause severe obstruction and lead to hydronephrosis and irreversible renal damage if left untreated. In our patient, creation of a tube was required to allow voiding from the normal anatomical location rather than the distal coronal level. Because the urethra was narrow, a dorsal plate incision was necessary to achieve both functional and cosmetic integrity, making TIPU the appropriate choice. In contrast, simple meatotomy would have left the meatus at the coronal level with a narrow downward-directed stream, resulting in inferior functional and cosmetic outcomes. Moreover, TIPU has been reported to carry lower complication rates than other urethroplasty techniques in distal cases, further supporting our decision.[5,7,9] According to the EAU Paediatric Urology Guidelines (2025), meatal stenosis occurs in 5–15% of cases after hypospadias repair, particularly when the meatus is reconstructed under tension. Although graft use may further reduce the risk of restenosis, TIPU alone remains an accepted and reliable option in distal cases, balancing functional durability with favorable cosmesis.[3,9,10]

EAU Paediatric Urology Guidelines (2025) stress the need for long-term follow-up in hypospadias patients, with periodic physical examination and uroflowmetry even in the absence of symptoms.[3,5,7] This case strongly supports these recommendations.

The presence of minimal bladder trabeculation on cystoscopy indicated chronic bladder outlet obstruction, despite the absence of other urethral strictures or posterior urethral valves. The discrepancy between bladder wall thickness on ultrasound and minimal trabeculation on cystoscopy may largely reflect differences in bladder filling volume at the time of assessment, as well as operator- and equipment-related variability. While ultrasound can overestimate thickness in a poorly filled or poorly compliant bladder, cystoscopy may underestimate subtle or focal trabeculation. The presence of minimal bladder trabeculation on cystoscopy indicated chronic bladder outlet obstruction, despite the absence of other urethral strictures or posterior urethral valves. Ultrasonographic assessment of bladder wall thickness is influenced not only by bladder filling status and compliance but also by the operator's experience, making it prone to inter-observer variability. In contrast, cystoscopy provides a direct visual evaluation but may underestimate subtle or focal changes. In our patient, longstanding obstruction likely resulted in muscular hypertrophy detectable by ultrasound, while cystoscopy revealed only mild mucosal changes. This highlights the need to interpret bladder findings in light of both imaging and endoscopic modalities. Although surgical correction restored normal voiding, bilateral renal scarring persisted, underscoring the consequences of delayed diagnosis.

CONCLUSION

Physical examination remains an indispensable, simple, and cost-effective diagnostic tool in pediatric urology. Distal urethral strictures can be easily missed if urinary symptoms are attributed solely to functional disorders. Routine meatal inspection and uroflowmetry should be part of the long-term follow-up after hypospadias repair, as timely recognition and intervention can prevent irreversible upper urinary tract damage.

ETHICAL DECLARATIONS

Informed Consent

Informed consent was obtained from the legal guardians of the pediatric patient described in this report. Where developmentally appropriate, assent was also sought from the child. The inclusion of vulnerable populations in this study adhered to national and international ethical guidelines. Extra care was taken to ensure voluntary participation, understanding, and protection of participant dignity and autonomy.

Peer Review Process

This report underwent external peer review.

Conflict of Interest

The authors declare no conflicts of interest.

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Author Contributions

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REFERENCES

1. Simonnet H, Lallemant-Dudek P. Urinary incontinence in children. *Rev Prat.* 2024;74(2):179-184.
2. Wu Y, Wang J, Zhao T, et al. Complications following primary repair of non-proximal hypospadias in children: a systematic review and meta-analysis. *Front Pediatr.* 2020;8:579364. doi:10.3389/fped.2020.579364
3. Radmayr C, Bogaert G, Bujons A, et al. EAU Guidelines on Paediatric Urology. Arnhem, The Netherlands: European Association of Urology; 2025. Available at: https://uroweb.org/guidelines/paediatric-urology. Accessed September 11, 2025.
4. González R, Ludwikowski BM. Importance of urinary flow studies after hypospadias repair: a systematic review. *Int J Urol.* 2011;18(11):757-761. doi:10.1111/j.1442-2042.2011.02839.x
5. Boroda J, Gitlin J, Fang A, et al. A comparison of 467 uroflowmetry results in repaired hypospadias vs. normal male flows. *J Pediatr Urol.* 2024;20(4):690.e1-690.e7. doi:10.1016/j.jpurol.2024.04.017
6. Andersson M, Doroszkiewicz M, Arfwidsson C, et al. Normalized urinary flow at puberty after tubularized incised plate urethroplasty for hypospadias in childhood. *J Urol.* 2015;194(5):1407-1413. doi:10.1016/j.juro.2015.06.072
7. Van der Horst H, De Wall L, Toelen J. Hypospadias, all there is to know. *Eur J Pediatr.* 2017;176(3):435-441. doi:10.1007/s00431-017-2864-5
8. Hueber PA, Antczak C, Abdo A, et al. Long-term functional outcomes of distal hypospadias repair: a single center retrospective comparative study of TIPs, Mathieu and MAGPI. *J Pediatr Urol.* 2015;11(2):68.e1-68.e7. doi:10.1016/j.jpurol.2014.09.011
9. Snodgrass WT, Bush N, Cost N. Tubularized incised plate hypospadias repair for distal hypospadias. *J Pediatr Urol.* 2010;6(4):408-413. doi:10.1016/j.jpurol.2009.09.010
10. Silay MS, 't Hoen L, Bhatt N, et al. Are there any benefits of using an inlay graft in the treatment of primary hypospadias in children? A systematic review and metanalysis. *J Pediatr Urol.* 2021;17(3):303-315. doi:10.1016/j.jpurol.2021.02.013

MHA MEDIHEALTH ACADEMY

Surgery on Children

Case Report

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Anesthesia management of an infant with laryngomalacia*

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ABSTRACT

Potential complications in the pediatric airway are significantly more frequent and diverse compared to adults. The underlying reasons include not only anatomical and physiological differences but also the higher prevalence of congenital anomalies affecting the airway in the pediatric population. Among these anomalies, the most common is laryngomalacia, characterized by the collapse of supraglottic structures during inspiration. Congenital laryngomalacia typically presents with stridor within the first 15 days of life, follows a self-limiting course, and tends to resolve by 24 months of age. In this report, we present our experience with difficult airway management and anesthesia in a 3.5-month-old, 5 kg patient diagnosed with laryngomalacia during surgery.

Keywords: Anesthesia, children, laryngomalacia, difficult airway

*This study was presented as an abstract at the International Health Sciences and Life Congress, held in Turkiye on April 16-19, 2015

INTRODUCTION

Laryngomalacia is the most common congenital laryngeal anomaly and represents the leading cause of stridor in neonates.[1] Although it may be present at birth, it typically manifests after the first four weeks of life with progressively worsening inspiratory stridor. The stridor is exacerbated by feeding, exertion, or the supine position, while it tends to improve with neck extension or prone positioning. Crying is usually normal, and no signs of lower respiratory tract involvement or cyanosis are observed. Clinical manifestations generally begin shortly after birth, increase in severity until the eighth month, plateau around the ninth month, and gradually resolve by 24 months of age.[2]

Pediatric difficult airway remains a major challenge for anesthesiologists and is recognized as one of the principal causes of perioperative respiratory complications.[3] A multicenter study conducted in the United States estimated the incidence of difficult airway in pediatric anesthesia to be between 2–5% per 1,000 cases. Moreover, approximately 20% of children requiring difficult airway management experienced complications.[4] In addition to the challenges encountered during endotracheal intubation, the limited time available for airway manipulation in children compared with adults further complicates management. Children diagnosed with laryngomalacia are therefore considered at increased risk for difficult airway management. In this report, we aim to present our experience in the anesthetic management of an infant with laryngomalacia.

CASE

Our patient was a 3.5-month-old male infant, weighing 5 kg and measuring 61 cm in length. He was referred to our department by the pediatric surgery clinic for preoperative evaluation prior to laparoscopic unilateral inguinal hernia repair and circumcision. His medical history revealed that he was born at 38 weeks of gestation with a birth weight of 1800 g and was diagnosed with low birth weight. Postnatally, he had persistent grunting respirations, for which he was evaluated by the pediatric clinic and diagnosed with laryngomalacia. Since no cyanosis or respiratory distress was observed at rest and oral intake was normal, close follow-up without medical intervention was recommended due to his mild symptoms.

Preoperative examinations and clinical evaluation revealed no signs of respiratory distress. According to the American Society of Anesthesiologists (ASA) classification, the patient was classified as ASA II. The infant, who was breastfed, was brought to the operating room after a 4-hour preoperative fasting period (**Figure 1**).

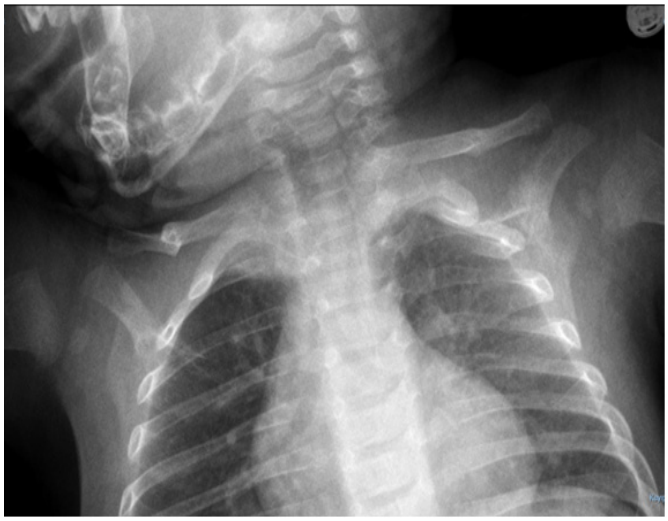

Figure 1. Chest X-ray

Anesthesia Management

Standard monitoring (heart rate, blood pressure, and SpO_2) was applied. Induction was initiated with 8% sevoflurane. After establishing peripheral intravenous access, 0.8 mcg/kg fentanyl and 1 mg/kg rocuronium were administered intravenously. Mask ventilation was easily achieved. Airway evaluation with a size 1 Miller laryngoscope revealed a difficult airway classified as Cormack–Lehane grade III. The first attempt at endotracheal intubation was unsuccessful; however, the patient was successfully intubated on the second attempt by an experienced anesthesiologist using a videolaryngoscope and a 3.0-mm uncuffed endotracheal tube. A 10 Fr orogastric tube was inserted.

The patient was then positioned laterally for caudal anesthesia. Following sterilization, a 25-gauge, 30-mm caudal needle was advanced through the sacral hiatus, and a caudal block was performed with 3 ml of 0.25% bupivacaine. Anesthesia was maintained with 2% sevoflurane in a 50% oxygen–air mixture. The surgical procedure lasted 67 minutes, during which intraoperative hemodynamic parameters remained stable.

For postoperative analgesia, in addition to the caudal block, 10 mg/kg intravenous paracetamol was administered. At emergence, 4 mg/kg sugammadex was given to reverse neuromuscular blockade, and the patient was successfully extubated without complications (**Figure 2**).

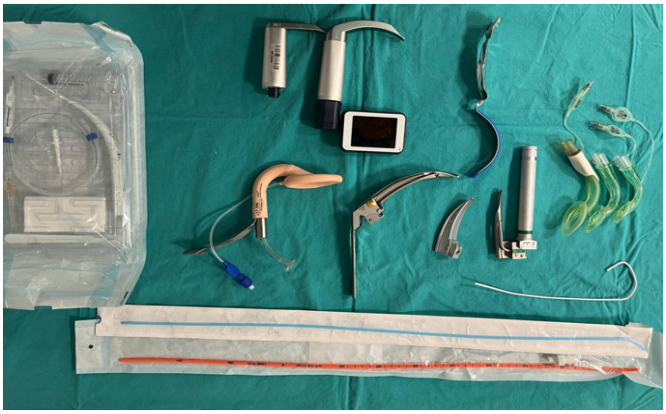

Figure 2. Preoperative airway preparation

Following surgery, the patient was transferred to the recovery room and closely monitored for approximately 40 minutes. As no signs of respiratory distress were observed, he was transferred to the pediatric surgery ward under nurse supervision.

DISCUSSION

Although the etiology of laryngomalacia has not yet been fully elucidated, several theories have been proposed regarding its pathogenesis. The most widely accepted hypothesis suggests that laryngomalacia results from inadequate neuromuscular maturation, which leads to supraglottic hypotonia and relaxation of these structures during inhalational anesthesia.[5]

Laryngomalacia is the most common cause of stridor in children, accounting for 65-75% of all cases.[6] It develops as a result of the inability to maintain laryngeal lumen patency during inspiration and occurs approximately twice as often in males compared with females. It is typically characterized by inspiratory stridor that spontaneously regresses within the first 24 months of life.[5] However, in approximately 10% of cases, severe airway obstruction may lead to marked dyspnea, dysphagia, growth retardation, and obstructive sleep apnea, requiring surgical intervention.[6] In our case, the patient was male and presented with inspiratory stridor, but did not exhibit symptoms severe enough to necessitate surgery (**Figure 1**).

The diagnosis of laryngomalacia is based on the clinical and epidemiological history of neonates and infants presenting with inspiratory stridor, which worsens with agitation or crying and improves during sleep. Direct laryngoscopy is considered the gold standard for definitive diagnosis.[7-9] In children with congenital or acquired airway pathology, airway management is of critical importance due to the risk of difficult intubation, difficult extubation, and the potential need for reintubation.[10] Children diagnosed with laryngomalacia are therefore at increased risk for difficult airway management. Most airway assessment methods and scoring systems validated in adults have not been confirmed in children. Nevertheless, children at risk for a difficult airway can be identified through a detailed medical history and comprehensive physical examination. In pediatric difficult airway scenarios, careful preparation and implementation of an appropriate anesthetic plan contribute to effective management and increase the likelihood of successful outcomes.[11]

A review of the literature highlights four main anesthetic techniques in the management of children with laryngomalacia and difficult airways: general anesthesia with intravenous maintenance, inhalational induction, high-frequency jet ventilation, and regional anesthesia (caudal or spinal) combined with sedation.[12] In our patient, general anesthesia was deemed necessary to ensure optimal surgical conditions, as the procedure was planned as a laparoscopic inguinal hernia repair.

When a difficult airway is anticipated, it is recommended to have advanced airway management techniques available, including videolaryngoscopy, supraglottic airway devices,

fiberoptic bronchoscopy, and needle cricothyrotomy. Placement of a laryngeal mask airway (LMA) has been reported to cause injuries to the uvula, pharyngeal mucosa, epiglottis, and other laryngeal structures.[13] These injuries may occur due to forceful insertion or as a result of downward folding of the epiglottis during placement. In our case, all necessary preparations—including readiness for emergency tracheostomy—were made, and the operation was initiated with full preparation for foreseeable risks (**Figure 2**). Following successful mask ventilation, difficult intubation was encountered; however, tracheal intubation was easily achieved on the second attempt using videolaryngoscopy with a smaller endotracheal tube. In cases of unexpectedly difficult tracheal intubation, repeated attempts should be avoided; instead, airway patency must be maintained, assistance sought, and alternative techniques capable of providing apneic oxygenation, along with appropriately sized intubation devices such as videolaryngoscopes, fiberoptic bronchoscopes, or supraglottic airway devices (with the use of a stylet or bougie if necessary), should be readily available. At this stage, tracheal intubation is recommended to be performed using alternative tools such as a videolaryngoscope or a fiberoptic bronchoscope.[14]

Videolaryngoscopy is particularly valuable in difficult airway management because of its potential to improve the success rate of tracheal intubation. Previous reports have demonstrated that the use of videolaryngoscopes significantly increases the first-pass success rate in children with difficult airways, including neonates and infants.[15] However, the effectiveness of videolaryngoscopy in small children may be less pronounced compared to that reported in older children and adults. Conversely, fiberoptic bronchoscope–guided intubation in neonates and infants with difficult airways has been shown to markedly increase first-pass success rates.[16] Nevertheless, it should be noted that fiberoptic bronchoscopes with an external diameter of <3 mm may not be readily available in every clinical setting.

In neonates with low birth weight, another important anesthetic goal is to minimize the risks of postoperative complications. In this patient population, the most frequent causes of postoperative airway obstruction include laryngospasm, laryngeal edema, and apnea, which is particularly common.[17] In our patient, all necessary preparations were undertaken, and the perioperative plan was meticulously executed. As a result of this cautious approach, no complications were encountered.

CONCLUSION

As a result, comprehensive preoperative evaluation and careful planning of anesthetic management are crucial in pediatric patients with anticipated difficult airways. Anticipating potential risks, implementing appropriate strategies, and ensuring the presence of an experienced anesthesiologist working in coordination with a skilled team play a key role in minimizing possible complications.

ETHICAL DECLARATIONS

Informed Consent

Informed consent was obtained from the legal guardians of the pediatric patient described in this report. Where developmentally appropriate, assent was also sought from the child. The inclusion of vulnerable populations in this study adhered to national and international ethical guidelines. Extra care was taken to ensure voluntary participation, understanding, and protection of participant dignity and autonomy.

Peer Review Process

This report underwent external peer review.

Conflict of Interest

The authors declare no conflicts of interest.

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Author Contributions

Concept: AD, CY, YAŞ; Design: AD, ESS; Control/Supervision: MK; Data Collection and/or Processing: AD, YAŞ; Analysis and/or Interpretation: CY, YAŞ; Literature Review: AD, ESS; Article Writing: AD, ESS, MK; Critical Review: All Authors.

REFERENCES

1. Jain D, Jain S. Management of stridor in severe laryngomalacia: a review article. *Cureus*. 2022;14(9):e29585. doi:10.7759/cureus.29585
2. Richter GT, Thompson DM. The surgical management of laryngomalacia. *Otolaryngol Clin North Am*. 2008;41(5):837-864. doi:10.1016/j.otc.2008.04.013
3. Cook TM, Woodall N, Frerk C. Major complications of airway management in the UK: results of the Fourth National Audit Project of the Royal College of Anaesthetists and the Difficult Airway Society. Part 1: anaesthesia. *Br J Anaesth*. 2011;106(5):617-631. doi:10.1093/bja/aer058
4. Fiadjoe JE, Nishisaki A, Jagannathan N, et al. Airway management complications in children with difficult tracheal intubation from the Pediatric Difficult Intubation (PeDI) registry: a prospective cohort analysis. *Lancet Respir Med*. 2016;4(1):37-48. doi:10.1016/S2213-2600(15)00508-1
5. Duprat AD, Campos CAH, Costa HOO. Tratado de otorrinolaringologia. 1st ed. Vol 4. São Paulo, Brazil: Roca; 2003:440-450.
6. Olney DR, Greinwald JH, Smith RJ, Bauman N. Laryngomalacia and its treatment. *Laryngoscope*. 1999;109(11):1770-1775. doi:10.1097/00005537-199911000-00011
7. Holinger LD. Diagnostic endoscopy of the pediatric airway. *Laryngoscope*. 1989;99(4):346-348. doi:10.1288/00005537-198904000-00011
8. Berkowitz RG. Neonatal upper airway assessment by awake flexible laryngoscopy. *Ann Otol Rhinol Laryngol*. 1998;107(1):75-80. doi:10.1177/000348949810700115
9. Moumoulidis I, Gray RF, Wilson T. Outpatient fiber-optic laryngoscopy for stridor in children and infants. *Eur Arch Otorhinolaryngol*. 2005;262(3):204-207. doi:10.1007/s00405-004-0804-3
10. Biricik E. Pediatric syndromes that may cause difficult airway. *Cukurova J Anesth Surg Sci*. 2023;6(2):366-374. doi:10.36516/cjass.1310065
11. Krishna SG, Bryant JF, Tobias JD. Management of the difficult airway in the pediatric patient. *J Pediatr Intensive Care*. 2018;7(3):115-125. doi:10.1055/s-0038-1624576
12. Patel N, Madi P, Monteiro I, Shah SP. The anesthetic management of a child with Ohtahara syndrome and severe stridor: a case report. *BMC Pediatr*. 2024;24(1):434. doi:10.1186/s12887-024-04907-8

13. Takeshita J, Nishiyama K, Fujii M, et al. Repetitive postoperative extubation failure and cardiac arrest due to laryngomalacia after general anesthesia in an elderly patient: a case report. *J Anesth*. 2017; 31(5):779-781. doi:10.1007/s00540-017-2373-8
14. Jagannathan N, Asai T. Difficult airway management: children are different from adults, and neonates are different from children! *Br J Anaesth*. 2021;126(6):1086-1088. doi:10.1016/j.bja.2021.03.012
15. Garcia-Marcinkiewicz AG, Kovatsis PG, Hunyady AI, et al. First-attempt success rate of video laryngoscopy in small infants (VISI): a multicentre, randomised controlled trial. *Lancet*. 2020;396(10266):1905-1913. doi:10.1016/S0140-6736(20)32532-0
16. Burjek NE, Nishisaki A, Fiadjoe JE, et al. Videolaryngoscopy versus fiber-optic intubation through a supraglottic airway in children with a difficult airway: an analysis from the multicenter pediatric difficult intubation registry. *Anesthesiology*. 2017;127(3):432-440. doi:10.1097/ALN.0000000000001758
17. Yeniay D, Değermenci M, Yucak Özdemir A. Laringomalazili bir yenidoğanda bilateral kasık fıtığı onarımı için kaudal anestezi. *Hitit Med J*. 2024;6(2):236-240. doi:10.52827/hititmedj.1444951

MHA MEDIHEALTH ACADEMY

Surgery on Children

Case Report

doi: 10.51271/SOC-0071

Congenital trifid tongue with macroglossia: a rare orofacial anomaly

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ABSTRACT

Trifid tongue is a rare congenital structural anomaly in which the anterior portion of the tongue is divided into three segments. It is most often associated with syndromic conditions such as orofacial digital syndrome. Neonates with a trifid tongue may present with airway obstruction, feeding difficulty, and drooling, necessitating early intervention. A 10-day-old full-term male neonate was admitted with feeding difficulty, drooling of saliva, choking episodes, and transient apnea. He had macroglossia and was unable to breastfeed. Surgical correction was performed using excision of the central tongue lobe and trimming of the medial aspects of the lateral lobes, followed by layered closure with absorbable sutures, preserving lingual neurovascular structures. The patient achieved effective sucking and feeding postoperatively. Early surgical management in congenital trifid tongue can result in excellent functional outcomes. A comprehensive evaluation to exclude associated syndromes is recommended.

Keywords: Trifid tongue, macroglossia, cleft soft palate, neonate, tongue reduction surgery

INTRODUCTION

Trifid tongue is a congenital and structural defect of the tongue in which its anterior end is divided longitudinally for a greater or lesser distance into three parts. The prevalence of trifid tongue associated with oro-facial digital syndrome type 1 is 1 in 50,000 to 1 in 250,000 live births.[1] But due to the rarity of the trifid tongue, the prevalence is not given in the literature. We present a rare case of trifid tongue accompanied by a soft cleft palate, with involvement of the anterior one-third of the tongue

Incomplete or abnormal development of structures derived from the lower half of the first branchial arch is rare; it may present as a complete or incomplete cleft of the lower lip, mandible, and tongue.[2]

The tongue is one of the main embryological structures that is derived from the pharyngeal apparatus. Towards. By the 5th to 6th gestational week, the (oral part) anterior 2/3rd develops from the fusion of two lateral lingual swellings and a median tongue bud called tuberculum impar.[3] The trifid tongue is formed by the failure of the fusion of lingual swellings with each other and with the tuberculum impar.[4]

A patient with an enlarged tongue may have symptoms of sleep apnea, respiratory distress, drooling, difficulty speaking, swallowing, and sucking. In the first two years of life, the airway symptoms in infants usually improve with supportive care. Urgent tongue reduction is recommended in infants with severe airway obstruction secondary to an enlarged tongue.[1]

CASE

A 10-day-old male baby weighing 3 kg was admitted to the pediatric ICU at Shaikh Zayed Hospital, Lahore. Examination at birth revealed an enlarged tongue. The patient was born at full term via cesarean section and passed meconium after 36 hours of his birth. He was unable to breastfeed because of an enlarged tongue since birth, and was being fed with a dropper.

He presented to the emergency department with complaints of transient apneic spells, though not quantified with SpO_2 values, choking, difficulty in sucking his mother's breast, drooling of saliva, constipation for 1 week, and abdominal distention for 1 day. On admission, he maintained an oxygen

saturation (SpO_2) of 94%-98% on room air. However, during episodes of apnea, transient desaturation was noted, with SpO_2 levels decreasing to approximately 88%–90%. In view of these recurrent apneic spells and airway compromise secondary to macroglossia, the patient was supported preoperatively with supplemental oxygen at 1 L/min via nasal cannula. There were no other antenatal and postnatal events. There was no history of any congenital anomaly in the family. He was the first child born to consanguineous parents.

On detailed examination after admission, the tongue was hypertrophied. Adequate examination of the soft and hard palate was not possible due to macroglossia. The mandible was normal, and the patient was unable to close his mouth due to an enlarged tongue. The abdomen was distended, with no palpable viscera. The spine and external genitalia were normal, and the anus was in its normal position. The remainder of the systemic examination was unremarkable. A workup for constipation was planned, along with examination of the oral cavity under anesthesia.

The patient was kept nil per os, and a microenema was given. He passed a small amount of stool, and abdominal distention was reduced. Complete blood count and serum biochemistry were within normal limits. A plain X-ray of the abdomen showed dilated bowel loops. Barium studies and ultrasound examination of the abdomen and pelvis were reported to be normal. The patient was operated on the 12th day of life, and a detailed examination of the oral cavity was done under general anesthesia; findings were noted, the tongue was hypertrophied, W-shaped, having three projections (**Figure 1**), and a soft cleft palate. The hard palate and mandible were normal. There was no evidence of ankyloglossia. Surgery was planned to correct the trifid tongue after detailed examination.

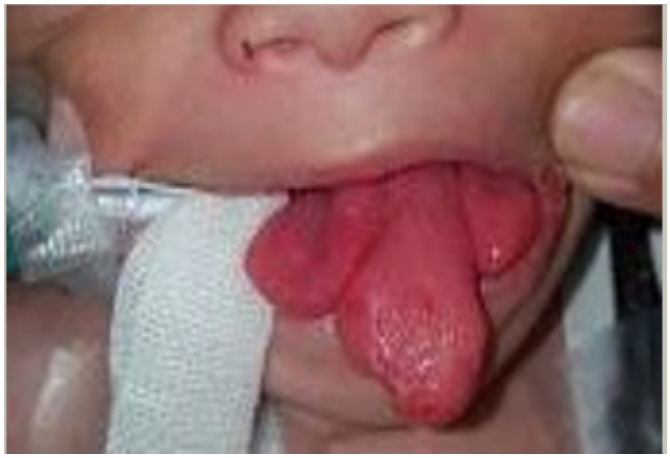

Figure 1. Preoperative intraoral photograph demonstrating a markedly hypertrophied tongue with a characteristic trifid configuration, showing three anterior lobulated projections consistent with congenital trifid tongue. The enlarged tongue occupied the oral cavity and contributed to impaired mouth closure and feeding difficulty.

Under general anesthesia, orotracheal intubation was achieved without difficulty, despite the enlarged tongue. The operative approach followed the principles of the Oji uniform tongue reduction technique, with minor modifications adapted to the trifid morphology.

Intraoperatively, the tongue was exposed using traction sutures placed at the anterior tip. A central wedge-shaped excision was designed along the midline, encompassing the entire central lobe of the trifid tongue. The excision extended from the anterior free margin posteriorly toward, but not beyond, the level of the circumvallate papillae. Cutting diathermy was used for excision to reduce tongue thickness. The resected segment measured approximately 2.0–2.5 cm in length and 1.0–1.2 cm in width at its widest anterior portion, tapering posteriorly. Additional controlled trimming of approximately 3–4 mm was performed along the medial edges of both lateral lobes to achieve symmetric contouring and adequate reduction in tongue width. Excision was limited to the intrinsic tongue musculature and performed using cutting diathermy to minimize blood loss. Particular care was taken to preserve the lingual arteries and the hypoglossal and lingual nerves by avoiding deep lateral dissection near the neurovascular bundles. Reconstruction was achieved with layered closure in three planes: approximation of the intrinsic musculature, closure of the submucosal layer to eliminate dead space, and mucosal closure using absorbable polyglactin sutures to restore a linear midline contour (**Figure 2, 3**).

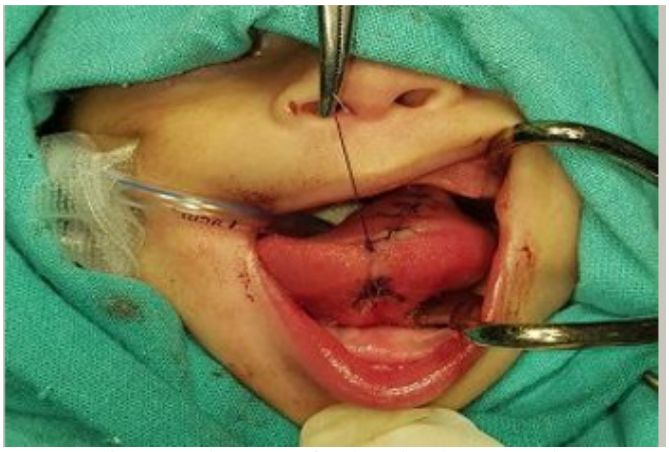

Figure 2. The image shows a uniformly reduced tongue with restoration of a linear midline contour, achieved after excision of the central lobe and controlled trimming of the medial aspects of the lateral lobes, consistent with a modified Oji tongue reduction technique

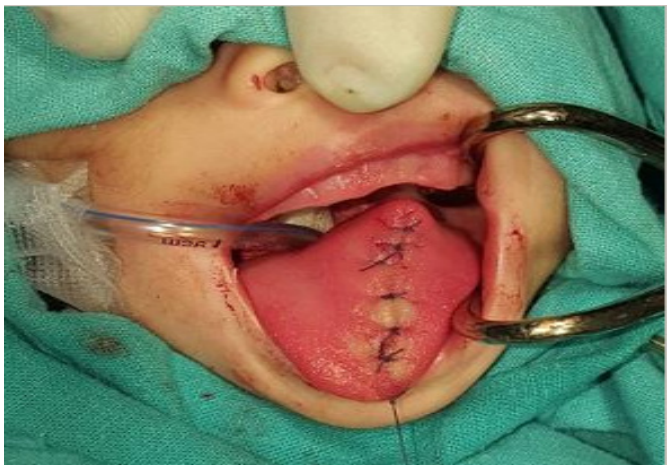

Figure 3. Immediate postoperative intraoral photograph demonstrating satisfactory tongue contour following reduction. The tongue shows reduced length and width, absence of lobulation, and adequate oral cavity space, allowing improved mouth closure. Sutured midline reconstruction is visible with preserved symmetry.

The surgery on the soft palate was deferred until the 3rd month of age. Excised tissue was sent for biopsy. Rectal biopsy was also done to rule out the suspicion of Hirschsprung's disease under the same anesthesia. The total operative time was approximately 40 minutes. There were no intraoperative complications, including bleeding, airway instability, or neurovascular injury. Postoperatively, the patient was maintained on prophylactic oxygen inhalation at 0.5 L/min, which was gradually tapered and discontinued on the first postoperative day without recurrence of desaturation episodes.

The patient demonstrated satisfactory tongue mobility, improved lip closure, and successful initiation of oral feeding by the third postoperative day with good sucking. Biopsy of the resected tissue showed normal tongue tissue. On rectal biopsy, ganglion cells and fine nerve fibrils were present. Ideally, genetic testing, detailed radiological studies, echocardiography, pulmonary function tests, CT scan, and ultrasound of the kidney, ureters, and bladder were indicated in this patient to rule out associations with other syndromes, but the parents refused these tests. Based on the results of the clinical and physical examinations, no additional anomalies were identified; however, the absence of advanced genetic and systemic assessments constitutes a limitation of this study.

DISCUSSION

Macroglossia with tongue lobulation has been described in association with several systemic conditions, including Beckwith–Wiedemann syndrome, vascular malformations, congenital hypothyroidism, glycogen storage disorders, and chromosomal abnormalities, most commonly Down syndrome.[1]

Trifid tongue is most frequently reported as part of the oro-facial-digital syndromes (OFDS), a heterogeneous group of disorders characterized by malformations of the oral cavity, craniofacial structures, and digits.[2] OFDS type I is the most common subtype, inherited in an X-linked dominant pattern and typically lethal in males, making live male presentation extremely uncommon.[3]

Mohr syndrome (OFDS type II), an autosomal recessive disorder, is characterized by a median cleft lip, a multilobed tongue, dental anomalies, and polydactyly.[5] However, most reported cases of trifid tongue occur in syndromic contexts rather than as isolated anomalies.

In our case, trifid tongue was associated with just a soft cleft palate and no evidence of facial dysmorphism, limb abnormalities, or systemic involvement.

Congenital tongue anomalies have also been reported in other syndromes such as Klippel–Feil syndrome and in infants of diabetic mothers, although these associations were not present in our case.[6,7]

Surgical intervention was indicated due to significant feeding difficulty, airway compromise, and inability to achieve adequate oral closure.

Unlike peripheral excision, which may leave a bulky central tongue, or wedge resection techniques that primarily reduce length while inadequately addressing tongue width, the Oji technique provides a balanced reduction in length, width, and thickness. Compared with the classic keyhole technique, the Oji method avoids creating a postoperative "square-shaped" tongue, which has been associated with adverse functional and speech outcomes as the child grows.[8-10]

The Oji tongue-reduction technique is a central-wedge glossectomy originally described for the management of diffuse macroglossia, most commonly in syndromic conditions such as Beckwith–Wiedemann syndrome. The classical indication of this technique involves diffuse tongue hypertrophy rather than focal or cleft-related malformations.

In our case, the indication was not macroglossia but congenital trifid tongue. Therefore, the Oji technique was modified to address the unique trifid morphology.

The principal modification consisted of tailoring the central wedge excision to incorporate the entire central lobe of the trifid tongue, rather than performing a symmetric volume reduction for diffuse macroglossia. A midline wedge was designed to excise the redundant central segment responsible for the trifid configuration. The excision extended from the anterior free margin posteriorly to just anterior to the circumvallate papillae, consistent with Oji principles of limiting posterior extension. However, unlike the classical Oji reduction aimed at global volumetric reduction, the resection in this case was anatomically corrective and morphology-driven.[2-11]

In our patient, tongue reduction using the Oji technique resulted in satisfactory functional and cosmetic outcomes, with early restoration of feeding and preserved tongue mobility. Histopathology confirmed normal tongue tissue.

CONCLUSION

Whenever a patient presents with such an anomaly, proper history, detailed examination, and detailed investigations should be performed to rule out associated anomalies. Genetic counseling for the family can help individuals make informed decisions about their medical condition. Molecular genetic testing enables confirmation of this disease and provides counseling to family members.

ETHICAL DECLARATIONS

Informed Consent

Informed consent was obtained from the legal guardians of the pediatric patient described in this report. Where developmentally appropriate, assent was also sought from the child. The inclusion of vulnerable populations in this study adhered to national and international ethical guidelines. Extra care was taken to ensure voluntary participation, understanding, and protection of participant dignity and autonomy.

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This report underwent external peer review.

Conflict of Interest

The authors declare no conflicts of interest.

Financial Disclosure

This case report did not receive any financial support.

Author Contributions

Concept: SI; Design: SI; Control: SI; Data Collection and/or Processing: AI; Analysis and/or Interpretation: AI; Literature Review: MAS; Article Writing: SI; Critical Review: All Authors.

REFERENCES

1. Coran AG, Caldamone A, Adzick NS, Krummel TM, Laberge J-M, Shamberger R. Pediatric Surgery E-book: Elsevier Health Sciences; 2012.
2. Oji C, Enyanwuma I, Henry G, Maduba C. The Oji technique: a new method of tongue reduction in a rare combination of multiple orofacial anomalies. *Oral Health Case Rep.* 2016;2(110):2. doi:10.4172/OHCR.1000110
3. Emmanouil-Nikoloussi EN, Krameos-Foroglou C. Developmental malformations of human tongue and associated syndromes. *Bull Group Int Rech Sci Stomatol Odontol.* 1992;35(1-2):5-12.
4. Ramanappa M, Reddy KJ. Foetal ultrasound–a study of spina bifida and associated anomalies in rural pregnant women. Indian Journal of Mednodent and Allied Sciences. 2016;4(2):59-63. doi:10.5958/2347-6206.2016.00009.1
5. Feather SA, Winyard PJ, Dodd S, Woolf AS. Oral-facial-digital syndrome type 1 is another dominant polycystic kidney disease: clinical, radiological and histopathological features of a new kindred. *Nephrol Dial Transplant*. 1997;12(7):1354-1361. doi:10.1093/ndt/12.7.1354
6. Öztürk F, Doruk C. Orthodontic treatment of a patient with oral-facial-digital syndrome. *Am J Orthod Dentofacial Orthop.* 2012;141(4 Suppl):S110-S118. doi:10.1016/j.ajodo.2010.09.033
7. Gillerot Y, Heimann M, Fourneau C, Verellen-Dumoulin C, Van Maldergem L. Oral-facial-digital syndrome type I in a newborn male. *Am J Med Genet*. 1993;46(3):335-338. doi:10.1002/ajmg.1320460318
8. Ferrante MI, Zullo A, Barra A, et al. Oral-facial-digital type I protein is required for primary cilia formation and left-right axis specification. *Nat Genet*. 2006;38(1):112-117. doi:10.1038/ng1684
9. Sakai N, Nakakita N, Yamazaki Y, Ui K, Uchinuma E. Oral-facial-digital syndrome type II (Mohr syndrome): Clinical and genetic manifestations. *J Craniofac Surg.* 2002;13(2):321-326. doi:10.1097/00001665-200203000-00028
10. Widgerow AD. Klippel-Feil anomaly, cleft palate, and bifid tongue. *Ann Plast Surg.* 1990;25(3):216-222. doi:10.1097/00000637-199009000-00014
11. James AW, Culver K, Hall B, Golabi M. Bifid tongue: a rare feature associated with infants of diabetic mother syndrome. *Am J Med Genet A*. 2007;143A(17):2035-2039. doi:10.1002/ajmg.a.31877

MHA MEDIHEALTH ACADEMY

Surgery on Children

Case Report

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Silent damage: asymptomatic jejuno-cecal fistula following multiple magnet ingestion in a child

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ABSTRACT

Multiple magnet ingestion in children constitutes a hazardous condition that may result in significant gastrointestinal complications. Sometimes, clinical symptoms may be entirely absent. This study seeks to demonstrate that multiple magnet ingestion can be asymptomatic clinically yet cause severe intestinal damage, and to underscore the importance of early surgical assessment for magnets that remain stationary on serial imaging, irrespective of the presence of symptoms. A seven-year-old male patient was evaluated one week following the ingestion of seven small spherical magnets. The patient remained asymptomatic, exhibiting no abdominal tenderness or signs indicative of peritonitis. Serial abdominal radiographs revealed persistent clustering of the magnets in the right lower quadrant, with no evidence of interval progression. Due to the lack of radiographic progression, a laparoscopic exploration was performed in the fifth week post-ingestion, following resolution of an intercurrent upper respiratory tract infection. A jejuno-cecal fistula resulting from pressure necrosis was identified approximately 45 centimeters distal to the ligament of Treitz. The affected intestinal segments were exteriorized via an enlarged umbilical incision. The magnets were extracted, the edges of the fistula were debrided, and the jejunal and cecal defects were primarily repaired in two layers. An appendectomy was also performed owing to the proximity of the cecal defect to the appendiceal base. No bowel resection was deemed necessary. The postoperative course was uneventful. This case illustrates that the ingestion of multiple magnets can result in severe intestinal injury, even in the absence of symptoms. Metallic foreign bodies that persist in a stationary position on serial imaging necessitate surgical exploration irrespective of symptom presentation.

Keywords: Children, foreign bodies, intestinal fistula, laparoscopy, magnets, play and playthings

INTRODUCTION

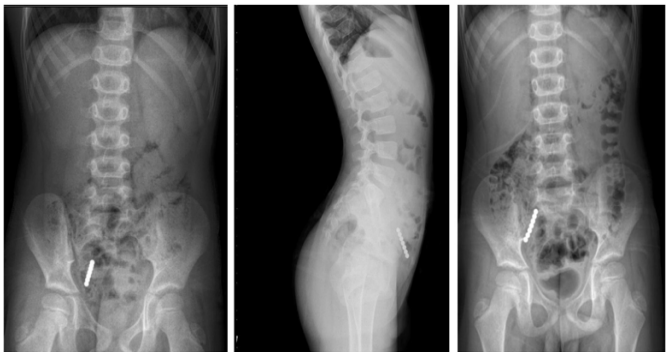
Foreign body ingestion constitutes a prevalent emergency within the pediatric demographic. Although the majority of foreign objects traverse the gastrointestinal tract without incident, the ingestion of multiple magnets at different instances presents a significant risk. Magnets possess the ability to attract each other through the intestinal wall, leading to their entrapment between segments of the intestine and resulting in tissue necrosis caused by compression, perforation, and fistula development.

Clinical manifestations may be minimal or entirely absent in cases where such complications arise. Consequently, there exists a potential for delays in both diagnosis and intervention. This report highlights a rare instance of a jejuno-cecal fistula that developed consequent to the ingestion of multiple magnets in a child who was entirely asymptomatic, along with an exploration of appropriate management strategies discussed.

CASE

A 7-year-old male patient with no known prior medical conditions was evaluated after reporting that he had swallowed seven small magnets following a bet with a friend. During the patient's presentation, it was learned that the incident occurred one week earlier. He had no complaints of abdominal pain, nausea, vomiting, fever, gastrointestinal bleeding, or changes in bowel habits (**Figure 1A**).

The patient was in good general condition. At presentation, his vital signs were stable: body temperature 36.4°C, heart rate 90 beats/min, respiratory rate 20 breaths/min, blood pressure



A B C

Figure 1. **A-B.** Abdominal A-P (**A**) and lateral (**B**) X-ray taken of the patient in a standing position at the time of admission to our clinic (7 days after swallowing). Demonstrating seven clustered spherical radiopaque magnets persistently localized in the right lower quadrant. **C.** Preoperative abdominal X-ray taken (30 days after swallowing). No change in the position of the contrast medium was observed.

110/70 mmHg, and oxygen saturation 98%. Initial laboratory evaluation showed no significant abnormalities. Abdominal examination was unremarkable, with normal bowel sounds and no signs of peritonitis or systemic infection.

One week after ingestion, the patient informed his family and presented to the hospital. An upright abdominal radiograph obtained on the day of presentation (day 7 after ingestion) demonstrated seven round radiopaque foreign bodies aligned in a linear configuration in the right lower quadrant/pelvic region. Because of the delayed presentation, the patient's asymptomatic status, and the clustered appearance of the magnets, the risks and benefits of conservative observation versus intervention were discussed with the family. Serial follow-up abdominal radiographs were obtained on the day of presentation and at 2, 7, 14, 18, 21, and 23 days after presentation (**Figure 1A, B**). (Corresponding to days 7, 9, 14, 21, 25, 28, and 30 after ingestion. On all radiographs, the magnets remained in the same right lower quadrant location and linear configuration, without separation or distal progression. In view of this persistent radiographic non-progression, surgical exploration was planned on the seventh day after presentation. However, because the patient had an active upper respiratory tract infection at that time, surgery was postponed until full clinical recovery for anaesthetic safety. As there were no signs of bowel obstruction or perforation and the patient remained entirely asymptomatic, close observation with serial radiographic follow-up was continued. After recovery from the upper respiratory tract infection, operative intervention was performed.

The patient underwent elective laparoscopic exploration. Intraoperatively, a fistulous communication was identified between the jejunum and cecum, approximately 45 cm distal to the ligament of Treitz, with magnets lodged across the tract. This caused pressure necrosis and localized erosion, establishing direct communication between the segments. No diffuse peritonitis or gross intra-abdominal contamination was observed. The umbilical camera-port incision was enlarged by approximately 2 cm, and the affected bowel loops were exteriorized. After careful separation, an opening in the jejunum was identified, providing direct access. Because the cecal defect was adjacent to the appendiceal base, appendectomy allowed removal of four magnets through the appendiceal lumen/stump, which avoided a separate cecal enterotomy. Finally, three magnets were removed directly through the jejunal opening (**Figure 2, 3**).

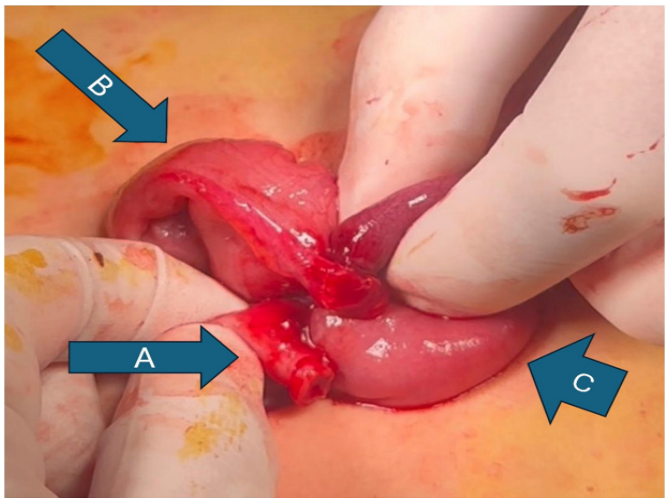


Figure 2. The jejuno-cecal anastomosis, which had formed a fistula, was brought out through the port inserted via the umbilicus under laparoscopic assistance. **A.** Post-appendectomy, **B.** Cecal loop, **C.** Jejunum loop adhered to the cecum with magnets

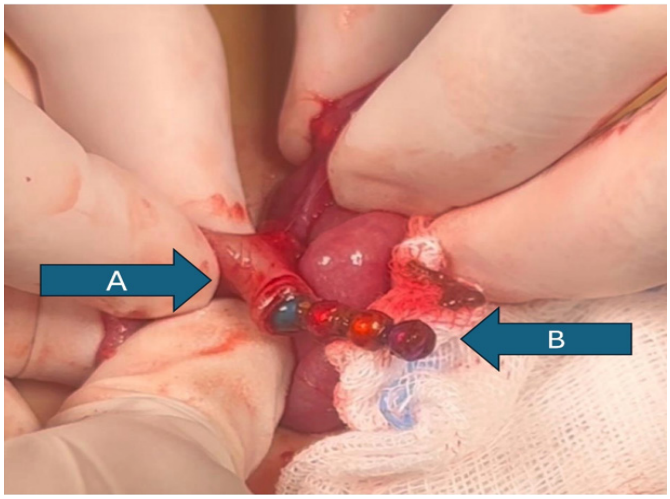


Figure 3. A. Removal of magnets from the cecum after appendectomy, B. Magnet

The fistula openings in both intestinal segments were debrided down to the limits of viable tissue and repaired primarily with absorbable sutures (4/0 90% glycolide and 10% L-lactide) in a double layer, thereby avoiding the necessity for resection. Due to the proximity of the cecal fistula opening to the appendix root, an appendectomy was performed. Intestinal viability was preserved, and no additional resection was required.

Follow-up

The patient's postoperative course was uneventful. He tolerated oral intake, bowel function returned to normal, and he was discharged on the third postoperative day with recommendations, without developing any postoperative complications. No problems were detected during outpatient follow-up visits.

DISCUSSION

Multiple magnet ingestion ranks among the conditions with the highest morbidity risk in pediatric foreign body ingestions. While a single magnet typically traverses the gastrointestinal tract without issue, the ingestion of multiple magnets at different times or their settlement in distinct intestinal segments can cause mutual attraction between intestinal loops. This interaction may result in compression-related ischemia, necrosis, perforation, and the formation of enteric fistulas.[1]

Although this pathological process generally manifests with symptoms during the acute phase, there may be delays in symptom onset or complete absence of symptoms. Guidelines and case series specifically highlight that compression necrosis can occur within hours, whereas gastrointestinal symptoms might not manifest until days or weeks later.[1] Consequently, the lack of clinical symptoms should not be regarded as a dependable indicator of a benign course.

A recent multicenter observational study demonstrated that, although most cases managed conservatively were asymptomatic, the complication rate was notably higher among the group necessitating surgical intervention.[2] Similarly, a systematic review published in 2025 reported that approximately 8% of patients who developed entero-enteric fistula due to multiple magnet ingestion were entirely asymptomatic.[3] These findings suggest that the potential for silent progression of severe intestinal damage should not be disregarded in clinical assessment.

In the scholarly literature, the lack of magnet displacement observed in serial radiographs is regarded as one of the most significant practical indicators necessitating surgical or endoscopic intervention. A multicenter study conducted in the United Kingdom found that 47.2% of 108 children who ingested multiple magnets required intervention, with the most prevalent reason being the lack of progression on serial imaging. The same study also reported intestinal perforation in 9.3% of cases and highlighted the importance of meticulous assessment, as "fixed" magnets imply that intestinal loops are adherent and interlinked.[4]

International guidelines also support early intervention. The ESGE/ESPGHAN (European Society of Gastrointestinal Endoscopy/The European Society for Pediatric Gastroenterology, Hepatology, and Nutrition) guidelines recommend removal of all endoscopically accessible magnets within 24 hours; for distal placements, they recommend close monitoring and surgical consultation if no progression is observed. The Royal College of Emergency Medicine (RCEM) and the North American Society for Pediatric Gastroenterology, Hepatology & Nutrition (NASPGHAN) algorithms also recommend frequent follow-up imaging, even in asymptomatic cases, and prompt intervention if no progression is evident.[1,6]

Delayed presentation or extended periods of conservative monitoring significantly elevate the risk of complications. A multicenter study conducted in 2024 reported a median interval from swallowing to presentation of 72 hours in the surgical group and 2 hours in the conservative group; the surgical cohort exhibited a perforation rate of 71.7%, an intensive care requirement of 20.8%, and a notably longer duration of hospitalization.[2] Furthermore, a single-center experience from China documented incidental detection of perforation 14 days later in an asymptomatic patient, with delayed presentation associated with an increased need for surgical intervention.[7] An 11-year cohort study from Turkiye also demonstrated that the presentation time was longer within the intervention group.[8]

The presented case exhibits two noteworthy features in the context of the existing literature. Firstly, the patient remained entirely asymptomatic despite the formation of a fistula. Although such cases are infrequently documented in systematic reviews, this subgroup holds significant clinical importance. Secondly, jejunocecal fistulas are exceedingly rare; within current reviews, this particular location has been documented in only isolated cases.[3]

In our case, the magnets remaining radiographically stable for 5 weeks, along with the small perforation size and the development of direct internal fistulization, explain the absence of peritonitis.[4] This finding is consistent with the localized damage mechanism described by Zheng et al.[7]

When these findings are evaluated collectively, it becomes evident that clinical symptoms do not constitute a dependable criterion for treatment decisions in cases of multiple magnet ingestion. Conversely, the absence of displacement of foreign bodies on serial imaging represents one of the most significant indicators of underlying severe intestinal damage.

CONCLUSION

The ingestion of multiple magnets constitutes a surgical emergency that can result in severe and potentially irreversible intestinal damage, even in the absence of clinical symptoms. The lack of movement of foreign bodies observed on serial radiographs indicates that the intestinal loops may be interlinked, and there may be occult perforation or fistulization. Consequently, regardless of symptom presentation, prompt surgical assessment and a proactive approach to exploration should be pursued in cases of multiple magnets without radiological evidence of progression.

ETHICAL DECLARATIONS

Informed Consent

Informed consent was obtained from the legal guardians of the pediatric patient described in this report. Where developmentally appropriate, assent was also sought from the child. The inclusion of vulnerable populations in this study adhered to national and international ethical guidelines. Extra care was taken to ensure voluntary participation, understanding, and protection of participant dignity and autonomy.

Peer Review Process

This report underwent external peer review.

Conflict of Interest

The authors declare no conflicts of interest.

Financial Disclosure

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Author Contributions

Concept/Design: GBB, SEÜB; Data Collection and/or Processing: OMÇ, BG, FBŞ; Analysis and/or Interpretation: OMÇ, MBÇ; Article Writing: OMÇ, GBB; Critical Review: GBB, OMÇ, SEÜB.

REFERENCES

1. Best Practice: Ingestion of Super Strong Magnets in Children - RCEM. Accessed January 8, 2026. https://rcem.ac.uk/best-practice/best-practice-ingestion-of-super-strong-magnets-in-children/
2. Alansari AN, Baykuziyev T, Soyer T, et al. Magnet ingestion in growing children: a multi-center observational study on single and multiple magnet incidents. *Sci Rep*. 2024;14(1):4575. doi:10.1038/s41598-024-55127-0
3. Kelaidi N, Dimopoulou K, Dimopoulou D, et al. Entero-enteric fistula following multiple magnet ingestion in children: a systematic review. *J Clin Med*. 2025;14(17):6235. doi:10.3390/JCM14176235/S1
4. Neville JJ, Harwood R, Bethell GS, et al. Magnet and button battery ingestion in children: multicentre observational study of management and outcomes. *BJS Open*. 2022;6(3):zrac056. doi:10.1093/BJSOPEN/ZRAC056
5. Tringali A, Thomson M, Dumonceau JM, et al. Pediatric gastrointestinal endoscopy: European Society of Gastrointestinal Endoscopy (ESGE) and European Society for Paediatric Gastroenterology Hepatology and Nutrition (ESPGHAN) Guideline Executive summary. *Endoscopy*. 2017;49(1):83-91. doi:10.1055/S-0042-111002
6. Kramer RE, Lerner DG, Lin T, et al. Management of ingested foreign bodies in children: a clinical report of the NASPGHAN Endoscopy Committee. 2015. doi:10.1097/MPG.0000000000000729
7. Zheng Y, Zhang Z, Yan K, et al. Retrospective analysis of pediatric patients with multiple rare-earth magnets ingestion: a single-center experience from China. *BMC Pediatr*. 2021;21(1):179. doi:10.1186/S12887-021-02642-Y
8. Varlıklı O, Akay MA, Sarıkaş NG, Metin S, Yıldız GE. The danger of magnet attraction: an 11-year cohort of pediatric intestinal complications due to magnet ingestion. *Turk J Pediatr*. 2025;67(5):720-731. doi:10.24953/TURKJPEDIATR.2025.6371

MHA MEDIHEALTH ACADEMY
Surgery on Children
Letter to the Editor
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"microRNAs (miRNAs) targeting oxidant-antioxidant mechanisms in primary ciliary dyskinesia and cystic fibrosis: epigenetic insights into therapeutic biomarkers"

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Dear Editor,

Primary ciliary dyskinesia (PCD) is a rare disorder marked by abnormalities in ciliary ultrastructure and/or function which are associated with disrupted mucociliary clearance and give rise to recurrent or persistent respiratory tract infections, bronchiectasis, bronchitis as well as a broad spectrum of clinical manifestations including sinonasal disease, neonatal respiratory distress, infertility, laterality defects and heterotaxy.[1]

Cystic fibrosis (CF) is also classified as a rare disease and is characterized by a defective CF transmembrane conductance regulator (CFTR) gene, whereas more than 40 disease-causing genes have been implicated in PCD to date.[2] CFTR dysfunction is associated with altered homeostasis in various organs and biological systems, including the pancreas, salivary glands, gastrointestinal tract, reproductive organs, and the respiratory system.[2,3] Within the framework of pulmonary homeostasis, impairment of mucociliary clearance constitutes a crucial pathogenic mechanism in CF which is linked to a sustained hyperinflammatory milieu and progressive pulmonary injury. Additionally, a close interconnection between inflammatory responses and the oxidant–antioxidant equilibrium was evident, as demonstrated by the significant correlations observed recently in our study.[4] Accordingly, levels of the intracellular antioxidant enzyme glutathione peroxidase (GPx) were significantly associated with the pro-inflammatory cytokines interleukin-1β (IL-1β), IL-6, and IL-8 in patients with PCD and CF.[4] From this perspective, the pro-inflammatory/-oxidant milieu and cellular antioxidant mechanisms represent pivotal targets for potential therapeutic strategies in patients with PCD and CF.

CF treatment is guided by symptomatic/supportive therapeutic modalities and novel therapeutic strategies including ion channel-targeted therapies, small-molecule approaches, and gene-based therapies.[2] Meanwhile, PCD exhibits a comparatively lower prevalence in relation to CF, notably among Caucasian populations and its clinical management continues to be predominantly guided by CF-derived therapeutic frameworks.[1,2]

It is noteworthy that microRNAs (miRNAs), a class of small non-coding RNAs, have garnered increasing attention in recent years as a potential therapeutic strategy in CF, due to their roles in modulating physiological functions and mediating disease-related pathophysiological mechanisms.[3] Among these miRNAs, miR-155 is regarded as a pivotal pro-inflammatory miRNA in patients with CF, with its increased expression being related with elevated IL-8 expression. In this context, miR-155-based therapeutic approaches can represent a promising strategy to restrict inflammatory responses by attenuating IL-8 mediated inflammatory processes.[5] Given the functional crosstalk between oxidative stress and inflammation, miRNAs associated with oxidant–antioxidant homeostasis have the potential to provide a relevant conceptual framework for research in both PCD and CF. Mitochondrial dynamics are well recognized as being indispensable for the maintenance of oxidant-antioxidant balance. In this context, miRNAs targeting genes associated with mitochondrial dynamics were assessed using bioinformatic approaches based on miRNA-target prediction tools, including TargetScan, Diana-microT and miRDB in this current study. Consequently, hsa-miR-369-3p, hsa-miR-381-3p and hsa-miR-382-3p were identified as candidate

miRNAs with the potential to regulate key mitochondrial dynamics related genes, including mitofusin 1 (MFN1), MFN2, and optic atrophy 1 (OPA1).

In conclusion, a comprehensive understanding of miRNAs as epigenetic regulators of genes related to mitochondrial dynamics reflecting oxidant-antioxidant equilibrium, particularly hsa-miR-369-3p, hsa-miR-381-3p and hsa-miR-382-3p, may provide critical insights to improve therapeutic outcomes and offer substantial translational relevance in these rare diseases.

ETHICAL DECLARATIONS

Peer Review Process

This letter was externally peer-reviewed.

Conflict of Interest

The author declare no conflicts of interest.

Financial Disclosure

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Author Contributions

The author is solely responsible for the entirety of conception, execution, analysis, and writing of the manuscript.

REFERENCES

1. Despotes KA, Zariwala MA, Davis SD, Ferkol TW. Primary ciliary dyskinesia: a clinical review. *Cells*. 2024;13(11):974. doi:10.3390/cells13110974
2. Pereira R, Barbosa T, Cardoso AL, Sá R, Sousa M. Cystic fibrosis and primary ciliary dyskinesia: similarities and differences. *Respir Med*. 2023;209:107169. doi:10.1016/j.rmed.2023.107169
3. Finotti A, Gambari R. Perspectives in microRNA therapeutics for cystic fibrosis. *Noncoding RNA*. 2025;11(1):3. doi:10.3390/ncrna11010003
4. Kartal Y, Bozdemir Özel C, Çakmak A, et al. The relationship between lung function, exercise capacity, oxidant and antioxidant response in primary ciliary dyskinesia and cystic fibrosis. *Turk J Pediatr*. 2024;66(3):309-322. doi:10.24953/turkjpediatr.2024.4581
5. Bhattacharyya S, Balakathiresan NS, Dalgard C, et al. Elevated miR-155 promotes inflammation in cystic fibrosis by driving hyperexpression of interleukin-8. *J Biol Chem*. 2011;286(13):11604-11615. doi:10.1074/jbc.M110.198390