

Causes and Consequences of Delayed Diagnosis of Anorectal Malformation in Children in a Resource-Constrained Environment

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Abstract: Delayed diagnosis of anorectal malformations (ARM) remains a frequent challenge in resource-constrained settings and is associated with increased neonatal morbidity and preventable complications. Evidence from low- and middle-income countries suggests that gaps in early neonatal examination and referral pathways contribute substantially to late presentation. **Objective:** To compare clinical presentation, early postoperative outcomes, and underlying causes of delayed diagnosis among neonates with anorectal malformations presenting to a tertiary care hospital in Pakistan.

Methods: A cross-sectional comparative study was conducted at Holy Family Hospital, Rawalpindi, Pakistan, from January 2024 to January 2025. Neonates with ARM were consecutively enrolled and categorized into early (<48 h of birth) and delayed (>48 h) diagnosis groups. Demographic characteristics, presenting features, postoperative complications following first-stage surgery, and reasons for delayed diagnosis were recorded. All patients underwent a standardized three-stage surgical protocol. Data were analyzed using SPSS v26.0. Continuous variables were compared using Student's t-test and categorical variables using Chi-square or Fisher's exact test. A p-value ≤ 0.05 was considered statistically significant. **Results:** A total of 82 neonates were included (41 per group). The mean age at presentation was significantly lower in the early diagnosis group (13 ± 8 h) compared with the delayed group (87 ± 25 h; $p < 0.01$). Abdominal distension (70.7% vs 26.8%; $p < 0.01$), vomiting (53.6% vs 22.0%; $p < 0.01$), and sepsis at admission (26.8% vs 4.8%; $p = 0.013$) were significantly more frequent among delayed cases. Urinary fistula and dehydration did not differ significantly between groups. Postoperative complications and early mortality were numerically higher in delayed cases but did not reach statistical significance. The most common reasons for delayed diagnosis were home delivery without a trained birth attendant, delayed referral from peripheral facilities, misdiagnosis, and absence of routine neonatal examination. **Conclusion:** Delayed diagnosis of ARM is associated with significantly worse clinical presentation, particularly higher rates of abdominal distension, vomiting, and sepsis at admission. Strengthening routine neonatal examination, improving early referral pathways, and promoting institutional deliveries may reduce diagnostic delays and improve early clinical outcomes in resource-limited settings.

Keywords: Anorectal malformation, complications, delayed diagnosis, causes, resource constraint

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Introduction

Anorectal malformations (ARMs) present significant surgical challenges and long-term health implications for children. Defined by defects in the formation of the anal and rectal structures, ARMs are among the most common congenital anomalies of the gastrointestinal system, with an estimated incidence ranging from 1 in 2,000 to 1 in 5,000 live births (1, 2). Timely and accurate diagnosis is essential, as delays in intervention not only exacerbate immediate medical complications but also have long-term consequences on morbidity and quality of life for affected children. Children with ARM may face difficulties with bowel control, psychosocial impacts, and increased healthcare resource utilization regardless of the healthcare context (3, 4).

Despite the availability of surgical correction techniques, delayed diagnosis remains prevalent in resource-constrained settings. Various studies indicate that such delays can occur due to limited access to healthcare services, insufficient awareness among healthcare providers, and inadequate diagnostic facilities. In a significant examination of pediatric surgical emergencies, it was found that many children in low-resource environments experience prolonged periods before diagnosis, resulting in increased rates of morbidity and mortality associated with these conditions (5, 2). For example, Saeed et al. highlighted that delayed diagnosis was linked to significant complications, including severe infections and prolonged hospitalization, in a cohort from a resource-limited pediatric ward (2).

In terms of specific metrics, a study focusing on ARMs revealed that up to 57% of cases were diagnosed later than recommended protocols, often after complications had developed (2). These findings are mirrored in other target conditions where healthcare access inequities manifest, reinforcing the critical need for improved diagnostic timelines and healthcare access strategies in low-income contexts (6, 7).

The challenges posed by delayed diagnosis are further exacerbated in specific demographics, such as in Pakistan, where healthcare systems often grapple with limited resources, inadequate training for healthcare professionals, and socio-cultural barriers that impede timely medical intervention. Pakistani children with ARMs face additional obstacles such as economic constraints, insufficient educational resources for healthcare awareness, and the cultural stigmatization of congenital conditions². These factors collectively contribute to a grim outlook for timely diagnosis and subsequent treatment outcomes.

The rationale for prioritizing research on delayed diagnosis in children with ARMs in resource-limited settings, particularly in countries like Pakistan, hinges on these multifaceted challenges that not only affect survival rates but also compromise the overall quality of life for affected children and their families. Addressing these issues is imperative for enhancing healthcare delivery frameworks, creating effective public health campaigns, and ultimately fostering an environment conducive to timely interventions (3, 8).

This study aims to ascertain the most prevailing consequences of delayed diagnosis of anorectal malformation in Rawalpindi District, Pakistan.



This will aid in improving the diagnosis and management of anorectal malformations, simultaneously providing us with a clearer understanding of our resource restrictions, leading to a delay in diagnosis.

Methodology

A cross-sectional comparative study was conducted at Holy Family Hospital, Rawalpindi, Pakistan, a tertiary-care government referral centre, over 13 months from January 2024 to January 2025. Ethical approval was obtained from the Institutional Review Board of Holy Family Hospital prior to study initiation. Written informed consent was obtained from the parents or legal guardians of all enrolled neonates. The study population comprised neonates diagnosed with anorectal malformations (ARM) who presented to the pediatric surgery unit during the study period.

Participants were categorized into two groups based on the timing of diagnosis. The early diagnosis group included neonates in whom ARM was identified during the initial neonatal examination or within the first 48 h of life. In contrast, the delayed diagnosis group comprised neonates presenting after 48 h of birth. Delayed diagnosis was operationally defined as failure to identify ARM within the first 48 h following delivery. All eligible patients were enrolled consecutively until the predetermined sample size was achieved.

The inclusion criteria consisted of all neonates with clinically confirmed ARM presenting during the study period. Patients who died prior to undergoing any surgical intervention and those managed with a single-stage procedure only, such as primary anoplasty without diversion and staged repair, were excluded to ensure homogeneity of surgical exposure and postoperative outcome assessment. Sample size estimation was performed using population-based parameters for Rawalpindi, with a 95% confidence level and 80% power, yielding a minimum sample size of 36 per group. Consecutive non-probability sampling was employed due to the low incidence of ARM and the tertiary referral nature of the study setting. The final sample exceeded the calculated minimum, strengthening the statistical precision of the comparisons.

Baseline demographic data and clinical characteristics at presentation, including age at diagnosis, sex, absence of anal opening, abdominal distension, vomiting, urinary fistula, dehydration, and sepsis at admission, were prospectively recorded using a standardized proforma. Postoperative outcomes following the first stage of surgical management, including wound dehiscence, wound infection, postoperative sepsis/septicemia, and in-hospital mortality, were documented through daily clinical assessments and review of medical records. Reasons for delayed diagnosis were systematically recorded through structured interviews with caregivers and review of referral documentation, including place of delivery, timing of referral, prior neonatal examination, and diagnostic accuracy at peripheral facilities, transport constraints, and sociocultural factors.

All enrolled neonates underwent a standardized three-stage surgical management protocol in accordance with institutional practice guidelines. The initial stage consisted of a diverting stoma to relieve intestinal obstruction and prevent fecal contamination. This was followed by definitive repair using posterior sagittal anorectoplasty after stabilization and optimization of the patient, and subsequently by stoma reversal once appropriate healing and functional assessment were achieved. Perioperative care, including antibiotic prophylaxis, fluid management, and postoperative wound care, was delivered in accordance with uniform departmental protocols to minimize treatment-related variability between groups.

Data were analyzed using SPSS version 26.0. Continuous variables were expressed as mean ± standard deviation and compared between groups using Student’s t-test after assessment of distributional assumptions. Categorical variables were summarized as frequencies and percentages and compared using the Chi-square test or Fisher’s exact test where appropriate. All statistical tests were two-tailed, and a p-value ≤0.05 was considered statistically significant.

Results

A total of 82 neonates with anorectal malformations were enrolled, exceeding the minimum calculated sample size. Patients were equally allocated to early diagnosis (<48 h) and delayed diagnosis (>48 h) groups (n = 41 each). The sex distribution was comparable between groups, with a slight male predominance that did not differ significantly (male: female 23:18 in the early group vs 22:19 in the delayed group). The mean age at presentation was significantly lower in the early diagnosis group (13 ± 8 h) than in the delayed diagnosis group (87 ± 25 h; p < 0.01). Absence of an anal opening and failure to pass meconium were observed in all patients. Clinical features at presentation differed between groups. Abdominal distension was significantly more frequent in delayed diagnosis (70.7%) compared with early diagnosis (26.8%; p < 0.01). Vomiting was also more common among delayed cases (53.6% vs 22.0%; p < 0.01). Sepsis at admission occurred in a higher proportion of neonates presenting late (26.8%) compared with those diagnosed early (4.8%; p = 0.013). The presence of urinary fistula, indicated by meconium in urine, did not differ significantly between the two groups (29.2% vs 19.5%; p = 0.44). Similarly, dehydration at presentation was more frequent in delayed cases but did not reach statistical significance (19.5% vs 7.3%; p = 0.19) (Table 1).

Postoperative outcomes following the first stage of surgical management were comparable between groups. Rates of wound dehiscence (12.2% vs 7.3%; p = 0.71), wound infection (19.5% vs 14.6%; p = 0.77), and postoperative sepsis/septicemia (4.8% vs 14.6%; p = 0.14) did not differ significantly between early and delayed diagnosis groups. Mortality occurred only among delayed cases (2 deaths; 4.8%), although this difference was not statistically significant (p = 0.15) (Table 2).

Among neonates with delayed diagnosis, the most frequently recorded reason for late presentation was home delivery without a trained birth attendant (n = 13), followed by late referral from peripheral healthcare facilities (n = 9), misdiagnosis by healthcare providers (n = 6), lack of routine neonatal examination at birth (n = 6), transport constraints (n = 4), and cultural hesitation in seeking care (n = 3) (Figure 1).

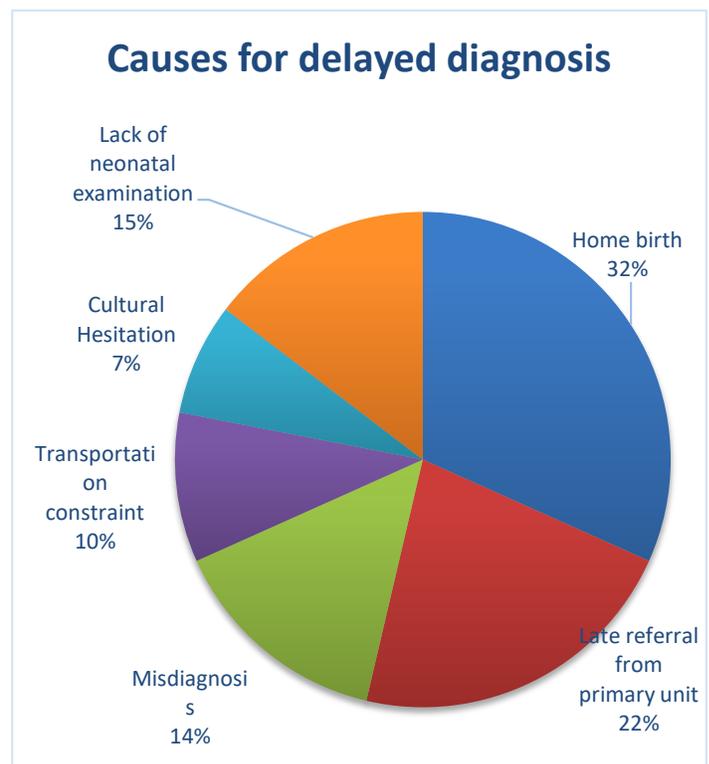


Figure 1: Causes of delayed diagnosis in the study population.

Table 1. Baseline characteristics and presenting features according to the timing of diagnosis

Parameter	Early Diagnosis (<48 h), n = 41	Delayed Diagnosis (>48 h), n = 41	p-value
Mean age at presentation (h), mean \pm SD	13 \pm 8	87 \pm 25	<0.01
Sex (male: female)	23:18	22:19	1.00
Absence of anal opening, n (%)	41 (100%)	41 (100%)	—
Abdominal distension, n (%)	11 (26.8%)	29 (70.7%)	<0.01
Vomiting, n (%)	9 (22.0%)	22 (53.6%)	<0.01
Urinary fistula (meconium in urine), n (%)	8 (19.5%)	12 (29.2%)	0.44
Sepsis at admission, n (%)	2 (4.8%)	11 (26.8%)	0.013
Dehydration, n (%)	3 (7.3%)	8 (19.5%)	0.19

Table 2. Postoperative outcomes following first-stage surgery

Outcome	Early Diagnosis (n = 41), n (%)	Delayed Diagnosis (n = 41), n (%)	p-value
Wound dehiscence	5 (12.2%)	3 (7.3%)	0.71
Wound infection	8 (19.5%)	6 (14.6%)	0.77
Sepsis/septicemia	2 (4.8%)	6 (14.6%)	0.14
Mortality	0 (0%)	2 (4.8%)	0.15

Discussion

The findings of our study delineating the effects of timing on the diagnosis of anorectal malformations (ARMs) among neonates illustrate critical patterns that align with the existing literature. A total of 82 neonates with ARMs were analyzed, resulting in two distinct groups: one consisting of patients diagnosed early (within 48 hours of birth) and the other, those diagnosed late (after 48 hours). The majority of existing studies corroborate our findings regarding the significant differences in clinical presentation and outcomes between these two cohorts.

The results indicate that the mean age at presentation was significantly lower in the early diagnosis group (13 \pm 8 h) than in the delayed diagnosis group (87 \pm 25 h; $p < 0.01$), underscoring the critical role of timely identification in minimizing complications (9, 10, 11). This is consistent with Chanchlani and Budhwani, who noted that early diagnosis was associated with improved overall outcomes in neonates with ARMs⁹. Similarly, Singh et al. illustrated that with delayed diagnosis, patients often present with more severe symptoms, which aligns with our findings that abdominal distension and vomiting occurred more frequently in delayed cases (70.7% vs. 26.8% and 53.6% vs. 22.0%, respectively; $p < 0.01$) (10).

Abdominal distension has previously been reported as a prevalent clinical sign in the context of delayed diagnosis and reflects the delayed metabolic and physiological deterioration faced by these neonates (12). Our study identifies sepsis at admission as a significant distinguishing factor between the two cohorts, occurring in 4.8% of early cases versus 26.8% of delayed ones ($p = 0.013$). This statistic reinforces the work of Mhammedi et al., who reported that delayed intervention often leads to critical clinical deterioration, thereby increasing the risk of sepsis among these patients (10, 12).

When examining postoperative outcomes, our study revealed no statistically significant differences in complications such as wound dehiscence, wound infection, or septicemia between early- and late-diagnosis groups (9, 11). This finding is noteworthy, as Abrahams et al. noted that while postoperative complications are common, early recognition often allows for better management strategies that can mitigate their severity (13). Our data indicating mortality was confined to the delayed diagnosis group (4.8%) further emphasizes the life-threatening nature of complications that can arise from late presentations (11).

The most frequently recorded reasons for late presentations among neonates with delayed diagnosis reflect broader systemic issues in healthcare access, with home deliveries without a trained birth attendant being cited as the primary factor ($n = 13$). This finding aligns with the study conducted by Perveen et al., which identified comparable barriers to timely care in Pakistan, including a lack of prenatal education and skilled healthcare personnel (14). The role of misdiagnosis and late

referrals, as noted in our study, corresponds to the findings by Sohail et al., who emphasized the challenge posed by inadequate training among primary healthcare providers in recognizing congenital anomalies (11).

This aligns with findings from recent literature, which highlight awareness and education among caregivers and healthcare providers as pivotal in reducing delays (15, 14). In resource-limited settings, a combined lack of healthcare resources and societal stigma regarding congenital conditions further complicates timely diagnosis and intervention (8, 16).

Our study corroborates existing literature that emphasizes the detrimental impact of delayed diagnosis on the clinical management of neonates with anorectal malformations. The significant differences in clinical presentation and outcomes between early- and late-diagnosis groups reveal critical areas for intervention within healthcare systems to improve awareness, access, and training for timely diagnosis and treatment. Understanding the barriers to care, particularly within the context of Pakistani healthcare, is essential in reducing the significant morbidity and mortality associated with the delayed diagnosis of such congenital anomalies.

Conclusion

Delayed diagnosis of ARM is associated with significantly worse clinical presentation, particularly higher rates of abdominal distension, vomiting, and sepsis at admission. Strengthening routine neonatal examination, improving early referral pathways, and promoting institutional deliveries may reduce diagnostic delays and improve early clinical outcomes in resource-limited settings.

Declarations

Data Availability statement

All data generated or analysed during the study are included in the manuscript.

Ethics approval and consent to participate

Approved by the department concerned. (IRBEC-HOLISB-551-24)

Consent for publication

Approved

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Conflict of interest

The authors declared the absence of a conflict of interest.

Author Contribution**NZ** (Resident)*Manuscript drafting, Study Design,***ZA** (Resident)*Review of Literature, Data entry, Data analysis, and drafting article.***BJ** (Resident)*Conception of Study, Development of Research Methodology Design,***HF** (Resident)*Study Design, manuscript review, critical input.***MFN** (Physiotherapist)*Manuscript drafting, Study Design,***IN** (Forensic scientist)*Review of Literature, Data entry, Data analysis, and drafting article.*

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