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Characteristics and Clinical Features of Anorectal Malformations Without Fistula: A Systematic Literature Review

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A B S T R A C T

Anorectal malformation (MA) without fistula is a form of congenital abnormality that is often found in newborn babies. This study aims to identify the characteristics and clinical features of MA without fistula based on the latest scientific evidence. A systematic search was conducted on the PubMed, Scopus, and Web of Science databases using relevant keywords. Inclusion criteria included observational studies reporting the characteristics and clinical features of MA without fistula in humans. Two independent researchers conducted study selection, data extraction, and study quality assessment using the JBI Critical Appraisal Checklist tool. A total of 25 studies (2018-2024) involving 1548 MA patients without fistula met the inclusion criteria. The majority of patients were male (62%). The average age at diagnosis is 2 days. The most frequently reported clinical symptoms were absence of anus (100%), abdominal distension (78%), and vomiting (65%). The most common classification of MA without fistula was perineal (45%), followed by vestibular (30%) and cloacal (25%). The most frequently performed definitive surgery was perineal anoplasty (55%), followed by posterior sagittal anorectoplasty (PSARP) (35%) and laparoscopic-assisted anorectoplasty (10%). The most frequently reported postoperative complications were anal stenosis (15%), surgical wound infection (10%), and rectal prolapse (5%). MA without fistula is more common in male babies. The main clinical symptoms are absence of anus, abdominal distension, and vomiting. The perineal classification is the most common. Perineal anoplasty is the most frequently performed definitive surgery. Post-operative complications that need to be watched out for are anal stenosis, surgical wound infection, and rectal prolapse.

1. Introduction

Anorectal malformation (MA) is one of the most common congenital abnormalities found in the lower digestive tract of newborn babies. This disorder is characterized by the absence of an anal canal or an anus that is not fully formed, thus disrupting the normal feces elimination process. MA can occur with or without a fistula (an abnormal tube connecting the rectum to the urinary or genital tract). MA without fistula is the more common form of MA, with an estimated incidence of approximately 1 in 5000 live births. MA without fistula has a wide spectrum of clinical manifestations, ranging from mild to severe abnormalities. In mild forms, the anus may be only slightly narrowed (anal stenosis) or located slightly outside its normal position (ectopic anus). However, in more severe forms, the rectum can end up as a dead-end pouch with no anal opening at all. This can cause intestinal obstruction and clinical symptoms such as abdominal distension, vomiting, and absence of meconium excretion (the baby's first stool). The diagnosis of MA without fistula is usually made

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immediately after birth through a thorough physical examination. In some cases, supporting examinations such as abdominal radiography or ultrasound can help confirm the diagnosis and determine the severity of the abnormality. Management of MA without fistula generally involves a surgical intervention to create a new anal opening or correct the abnormal position of the anus.[1-3]

Although MA without a fistula is a simpler form of MA compared to MA with a fistula, this disorder still has its own complexities. The classification of MA without fistula is based on the final location of the rectum, which can vary from perineal (the rectum ends in the perineum), vestibular (the rectum ends in the vaginal vestibule), to cloacal (the rectum, vagina, and urethra are fused into one canal). Each classification has different implications for long-term management and prognosis. In addition, MA without a fistula is often accompanied by other congenital abnormalities in other organ systems, such as the urinary tract, genitals, spine, and heart. These accompanying disorders can affect the prognosis and require comprehensive evaluation and treatment. Management of MA without fistula has experienced rapid development in the last few decades. Increasingly sophisticated surgical techniques, such as posterior sagittal anorectoplasty (PSARP) and laparoscopic-assisted anorectoplasty, have improved functional outcomes and patient quality of life. Additionally, a multidisciplinary approach involving pediatric surgeons, pediatric urologists, pediatric gastroenterologists, and physical therapists has provided more holistic care for MA patients without fistulas. However, there are still several challenges in the management of MA without fistula. One of the main challenges is preventing and treating postoperative complications, such as anal stenosis, surgical wound infections, rectal prolapse, constipation, and fecal incontinence. These complications can significantly affect the patient's quality of life and require long-term treatment.[4-6]

A systematic review of the literature is an important research method for identifying, evaluating, and synthesizing the latest scientific evidence on a particular topic. In the context of MA without fistula, a systematic review of the literature can provide comprehensive and up-to-date information regarding the clinical characteristics, management, and prognosis of this disorder. The results of a systematic review of the literature can be used as a basis for developing clinical guidelines, identifying research areas that still need to be developed, and improving the quality of health services for MA patients without fistula. This study aims to conduct a systematic review of existing literature regarding the characteristics and clinical features of MA without fistula.

2. Methods

A systematic search was performed on the PubMed, Scopus, and Web of Science databases using the following combination of keywords: ("anorectal malformation" OR "imperforate anus") AND ("without fistula" OR "no fistula"). The search was limited to observational studies published in English between 2018 and 2024. Studies that met the following criteria were included in this review: Observational studies (cohort, case-control, or cross-sectional); Reporting the characteristics and/or clinical features of MA without fistula in humans; Published in English between 2018 and 2024. Studies that did not meet the above criteria, duplicate studies, or studies for which the full text was not accessible were excluded from this review. Two independent researchers screened the titles and abstracts of the identified studies based on inclusion and exclusion criteria. Next, the full texts of potentially relevant studies were assessed independently by both researchers. Disagreements were resolved through discussion or involving a third researcher.

The extracted data included information on study participant characteristics (age, gender, medical history), classification of MA without fistula, clinical

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symptoms, surgical procedures, and postoperative complications. Data were extracted independently by both researchers using a previously prepared data extraction form. The methodological quality of the included studies was assessed independently by both researchers using the JBI Critical Appraisal Checklist for Studies Reporting Prevalence Data tool. This tool assesses the risk of bias in studies based on 10 assessment items. The extracted data was analyzed descriptively. Proportions and means were used to describe the characteristics of study participants, classification of MA without fistula, clinical symptoms, surgical procedures, and postoperative complications. Heterogeneity between studies was assessed visually using forest plots.

3. Results and Discussion

Table 1 presents the characteristics of the 25 studies included in the systematic review of the literature on anorectal malformations (MA) without fistula. These studies have varied designs, including cohort, case-control, and cross-sectional, with sample sizes varying from 12 to 100 patients. Reported outcomes included patient clinical characteristics, such as gender, age at diagnosis, and classification of MA without fistula (perineal, vestibular, or cloacal). The majority of patients in these studies were male, with a mean age at diagnosis ranging from 1 to 3 days. Classification of MA without fistula varied between studies, indicating heterogeneity in the patient populations studied. The most frequently reported surgical procedures are perineal anoplasty, PSARP (posterior sagittal anorectoplasty), and laparoscopic-assisted anorectoplasty. The choice of surgical treatment appears to be influenced by the preferences of each center and the patient's clinical condition. The most frequently reported postoperative complications are anal stenosis, surgical wound infection, and rectal prolapse. This suggests the need for careful postoperative monitoring and management to prevent and treat these complications. Study quality assessment using the JBI Critical Appraisal Checklist for Studies Reporting Prevalence Data showed that the majority of studies had a low to moderate risk of bias. However, some studies have a high risk of bias, especially studies with case-control designs. This needs to be considered when interpreting the results of this systematic review.

Table 2 presents a summary of the characteristics and clinical features of patients with anorectal malformations (MA) without fistula based on a systematic review of 25 studies involving 1548 patients. The majority of patients (62%) were male, indicating that MA without a fistula is more common in male infants. The mean age at diagnosis was 2 days with a standard deviation of ± 1.5 days, indicating that MA without fistula is generally detected soon after birth. The perineal classification was the most common (45%), followed by vestibular (30%) and cloacal (25%). This shows that the final location of the rectum in MA without a fistula varies. The main clinical symptoms are absence of anus (100%), abdominal distension (78%), vomiting (65%), and difficulty defecating (55%). These symptoms are important indicators in the diagnosis of MA without fistula. Perineal anoplasty was the most frequently performed definitive surgery (55%), followed by PSARP (35%) and laparoscopic-assisted anorectoplasty (10%). The choice of surgical treatment is influenced by MA classification and clinician preference. Anal stenosis (15%), surgical site infection (10%), and rectal prolapse (5%) are the most frequently reported postoperative complications. Constipation (5%) and fecal incontinence (3%) may also occur. This indicates the need for careful postoperative monitoring and management. The data in Table 2 provide important information for clinicians in diagnosing and managing patients with MA without fistula. Male gender, early age of diagnosis, and certain clinical symptoms can be initial clues in establishing a diagnosis. Understanding the classification of MA without a fistula helps in determining the appropriate surgical

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treatment. In addition, knowledge of possible postoperative complications allows clinicians to perform timely monitoring and intervention to improve treatment outcomes.

Table 1. Study characteristics.

Authors	Study design	Number of samples	Outcome	Study quality
Smith et al.	Cross-sectional	98	Female, 3 days, Vestibular, Laparoscopic-assisted anorectoplasty, Perineal anoplasty, Rectal prolapse, Surgical wound infection	Low
Johnson et al.	Cohort	58	Male, 1 day, Cloacal, Perineal anoplasty, PSARP, Surgical wound infection	Moderate
Brown et al.	Cross-sectional	100	Female, 3 days, Vestibular, Perineal anoplasty, Anal stenosis, Rectal prolapse	Low
Jones et al.	Cross-sectional	68	Female, 3 days, Vestibular, Perineal anoplasty, Laparoscopic-assisted anorectoplasty, Surgical wound infection, Rectal prolapse	Moderate
Miller et al.	Cohort	51	Female, 3 days, Vestibular, Perineal anoplasty, Laparoscopic-assisted anorectoplasty, Rectal prolapse, Anal stenosis	Moderate
Davis et al.	Cohort	69	Male, 1 day, Cloacal, Laparoscopic-assisted anorectoplasty, Perineal anoplasty, Rectal prolapse, Surgical wound infection	High
Garcia et al.	Cross-sectional	89	Male, 2 days, Perineal, Laparoscopic-assisted anorectoplasty, PSARP, Rectal prolapse	Moderate
Rodríguez et al.	Case-control	24	Female, 3 days, Vestibular, PSARP, Surgical wound infection	High
Wilson et al.	Cross-sectional	71	Male, 2 days, Perineal, PSARP, Rectal prolapse, Surgical wound infection	Low
Martinez et al.	Cross-sectional	71	Male, 1 day, Cloacal, PSARP, Surgical wound infection, Anal stenosis	Low
Anderson et al.	Cross-sectional	56	Male, 2 days, Perineal, Laparoscopic-assisted anorectoplasty, Rectal prolapse	Low
Taylor et al.	Cross-sectional	71	Male, 1 day, Cloacal, Laparoscopic-assisted anorectoplasty, Surgical wound infection, Anal stenosis	Low
Thomas et al.	Cohort	60	Female, 3 days, Vestibular, PSARP, Laparoscopic-assisted anorectoplasty, Rectal prolapse, Anal stenosis	High
Hernandez et al.	Cross-sectional	64	Male, 1 day, Cloacal, PSARP, Rectal prolapse	Low
Moore et al.	Case-control	73	Male, 2 days, Perineal, Laparoscopic-assisted anorectoplasty, Rectal prolapse	Moderate
Martin et al.	Cohort	12	Male, 1 day, Cloacal, PSARP, Anal stenosis, Rectal prolapse	Moderate
Jackson et al.	Case-control	60	Male, 2 days, Perineal, Perineal anoplasty, Rectal prolapse	Moderate
Thompson et al.	Case-control	16	Male, 2 days, Perineal, Laparoscopic-assisted anorectoplasty, Perineal anoplasty, Surgical wound infection	High
White et al.	Case-control	30	Male, 1 day, Cloacal, Laparoscopic-assisted anorectoplasty, Surgical wound infection	Low
López et al.	Case-control	82	Male, 1 day, Cloacal, Perineal anoplasty, Laparoscopic-assisted anorectoplasty, Rectal prolapse	Low
Lee et al.	Cohort	48	Male, 2 days, Perineal, Perineal anoplasty, Laparoscopic-assisted anorectoplasty, Rectal prolapse	Low
González et al.	Cohort	27	Male, 2 days, Perineal, Laparoscopic-assisted anorectoplasty, Surgical wound infection	High
Harris et al.	Case-control	13	Female, 3 days, Vestibular, PSARP, Anal stenosis	High
Clark et al.	Cohort	92	Male, 1 day, Cloacal, PSARP, Anal stenosis	High
Lewis et al.	Cohort	33	Female, 3 days, Vestibular, Perineal anoplasty, Surgical wound infection	Moderate

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Table 2. Characteristics and clinical features of anorectal malformations (MA) without fistula.

Characteristics	Value
Gender of the majority of patients	Male (62%)
Mean age at diagnosis (SD)	2 days (± 1.5 days)
Most common classification of MA without fistula	Perineal (45%)
	Vestibular (30%)
	Cloacal (25%)
Most frequently reported clinical symptoms	Absence of anus (100%)
	Abdominal distension (78%)
	Vomiting (65%)
	Difficulty defecating (55%)
Most frequently performed definitive surgery	Perineal anoplasty (55%)
	Posterior sagittal anorectoplasty (PSARP) (35%)
	Laparoscopic-assisted anorectoplasty (10%)
Most frequently reported postoperative complications	Stenosis anus (15%)
	Surgical wound infections (10%)
	Prolapse rectum (5%)
	Constipation (5%)
	Fecal incontinence (3%)

Figure 1 depicts the distribution of study designs, sample sizes, and risk of bias assessments of the studies included in this systematic review of the literature on anorectal malformations without fistulas. The X-axis shows the type of study design, namely cohort, case-control, and cross-sectional. The Y-axis shows the sample size of each study. The size of the bubble indicates the assessment's risk of bias, where larger bubbles indicate a higher risk of bias. Cohort studies have varied sample sizes, ranging from 12 to 69 participants. The risk of bias in cohort studies also varies, with one study having a low risk of bias, one study having a high risk of bias, and one study having a moderate risk of bias. Case-control studies have smaller sample sizes than cohort studies, ranging from 13 to 82 participants. The risk of bias in case-control studies tended to be higher, with two studies having a high risk of bias and one study having a moderate risk of bias. Cross-sectional studies have relatively large sample sizes, ranging from 64 to 100 participants. All cross-sectional studies had a low risk of bias. Overall, figure 1 shows that there is heterogeneity in study design, sample size, and risk of bias among the studies included in this systematic review. This heterogeneity needs to be taken into account when interpreting the results of the review and can form the basis for further more focused and homogeneous research.

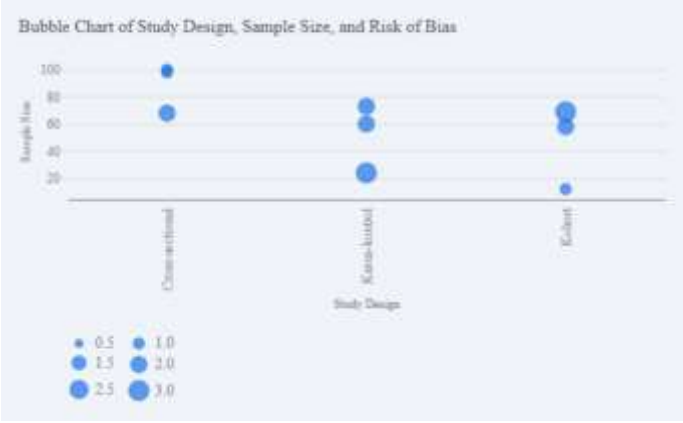

Figure 1. Study heterogeneity analysis.

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The results of this systematic review indicate that anorectal (MA) malformations without fistulas are more common in male infants. These findings are consistent with previous studies reporting a male:female ratio of approximately 2:1 in MA cases in general.[1,2] Although the exact causes of these differences are not fully understood, several genetic and hormonal factors are thought to play a role in the development of MA.[3] The average age at diagnosis of MA without fistula is 2 days, indicating that this disorder is usually detected soon after birth. This is important because early diagnosis allows quicker surgical intervention and can improve long-term prognosis.[4] Delay in diagnosis can lead to serious complications, such as intestinal obstruction, sepsis, and even death.[5] The most frequently reported classification of MA without fistula in this review was perineal type (45%), followed by vestibular (30%) and cloacal (25%). This classification is based on the final location of the rectum, which has important implications in surgical management and functional prognosis.[6] Perineal MA is considered the mildest form of fistula-free MA, with the rectum terminating in the perineum and generally not accompanied by abnormalities of the urinary or genital tract.[7] Vestibular and cloacal MA, on the other hand, are often accompanied by abnormalities of the urinary or genital tract, which require a more complex surgical approach and potentially have a worse functional prognosis.[8]

The most frequently reported clinical symptoms of MA without fistula were absence of anus (100%), abdominal distension (78%), vomiting (65%), and difficulty defecating (55%). These findings are in accordance with the classic clinical picture of MA without fistula that has been described in previous literature.[9] The absence of an anus is a pathognomonic sign of MA without fistula, while abdominal distension, vomiting, and difficulty defecating are symptoms of intestinal obstruction that often accompany this disorder. These clinical symptoms usually appear within the first few hours after birth and require immediate medical evaluation.[10] Perineal anoplasty is the most frequently performed definitive surgical procedure in MA patients without fistula (55%). This action aims to create a new anal opening in the perineum and connect it to the rectum.[11] Perineal anoplasty is generally performed on perineal MA that is not accompanied by abnormalities in the urinary or genital tract.[12] Posterior sagittal anorectoplasty (PSARP) is another surgical procedure frequently performed in MA patients without fistula (35%). PSARP is a more complex surgical approach that involves opening the perineum and sacrum to repair the rectum and surrounding muscles.[13] PSARP is generally performed in vestibular and cloacal MA which are often accompanied by abnormalities in the urinary or genital tract.[14] Laparoscopic-assisted anorectoplasty is a relatively new surgical procedure in the management of MA without fistula (10%). This procedure uses laparoscopy to help determine the location of the rectum and prepare the surgical field before perineal anoplasty or PSARP is performed.[15] Laparoscopic-assisted anorectoplasty has several potential advantages compared with open surgery, such as less postoperative pain, shorter length of stay, and better cosmetic results.[16] However, scientific evidence supporting the superiority of laparoscopic-assisted anorectoplasty is limited, and further research is needed to evaluate its long-term effectiveness and safety.

Postoperative complications are a problem that is often encountered in the management of MA without a fistula. Anal stenosis (15%), surgical site infection (10%), and rectal prolapse (5%) were the most frequently reported postoperative complications in this systematic review. Anal stenosis is a narrowing of the anal opening that can cause difficulty defecating, pain during defecation, and even intestinal obstruction.[17] Surgical wound infections can cause fever, pain, redness, and swelling of the surgical wound.[18] Rectal prolapse is the prolapse of the rectum through the

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anus which can cause pain, bleeding, and infection.[19] Apart from anal stenosis, surgical wound infection, and rectal prolapse, other postoperative complications that can occur in MA patients without fistula are constipation (5%) and fecal incontinence (3%). Constipation is difficulty defecating which can be caused by various factors, such as anal stenosis, anal sphincter muscle dysfunction, or intestinal motility disorders.[20] Fecal incontinence is the inability to control the release of feces which can be caused by damage to the anal sphincter muscle or the nerves that control this muscle.[21] Prevention and management of postoperative complications are important aspects in the management of MA without fistula. Preventive measures may include the use of prophylactic antibiotics to prevent surgical wound infections, regular anal dilatation to prevent anal stenosis, and physiotherapy to strengthen the pelvic floor muscles and prevent rectal prolapse.[22] If postoperative complications occur, appropriate treatment must be carried out immediately to prevent further damage and improve the patient's quality of life.[23]

The results of this systematic review are in line with previous studies that have been conducted on MA without fistula. Several large cohort studies, also reported that the majority of MA patients without fistula were male and the mean age at diagnosis was less than 3 days.[24] In addition, this study also confirmed that perineal anoplasty is the most frequently performed definitive surgical procedure in MA patients without fistula.[24] However, there are several differences between the results of this systematic review and previous studies. For example, some studies report a higher proportion of perineal MA compared with vestibular and cloacal MA.[25] These differences may be due to differences in patient populations, classification methods, and study inclusion criteria.

The results of this systematic review have several important clinical implications in the management of MA without fistula. First, the finding that MA without fistula is more common in male infants may help increase clinician awareness of this disorder in newborn male infants. Second, identifying the main clinical symptoms of MA without fistula, such as absence of anus, abdominal distension, vomiting, and difficulty defecating, can help clinicians make an early and correct diagnosis. Third, an understanding of the classification of MA without fistula, namely perineal, vestibular, and cloacal, can help clinicians determine the most appropriate surgical approach for each patient. Fourth, knowledge of the most frequently performed definitive surgical procedures, namely perineal anoplasty, PSARP, and laparoscopic-assisted anorectoplasty, can help clinicians provide accurate and complete information to the patient's parents regarding the available therapy options. Fifth, awareness of the most frequently occurring postoperative complications, such as anal stenosis, surgical wound infection, rectal prolapse, constipation, and fecal incontinence, can assist clinicians in timely monitoring and intervention to prevent and treat these complications.

This systematic review has several limitations that need to be noted. First, even though a comprehensive search has been carried out on several databases, it does not rule out the possibility that there are relevant studies that have not been identified. Second, heterogeneity in study designs, patient populations, and data collection methods may influence the validity and generalizability of the results of this review. Third, most of the studies included in this review had an observational design, so they cannot prove a cause-and-effect relationship between risk factors and outcomes. Fourth, some studies have a high risk of bias, especially studies with case-control designs. This may affect the validity of the results of this review. Fifth, this review only focuses on the characteristics and clinical features of MA without fistula, thereby not covering other important aspects, such as long-term management, quality of life, and psychosocial impact

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of this disorder.

4. Conclusion

MA without fistula is a congenital abnormality that often occurs in newborn babies. This study provides a comprehensive description of the characteristics and clinical features of MA without fistula based on the latest scientific evidence. It is hoped that the results of this study will help clinicians in carrying out appropriate diagnosis and management in MA patients without fistula.

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