*IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)*
*e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 20, Issue 7 Ser.13 (July. 2021), PP 13-15*
*www.iosrjournals.org*

# Low Anorectal Malformation in Males with Delayed Presentation – Issue of a Developing Country

Dr . Jay Kishor Soren, Dr. Priya Shalini Lakra*
*Corresponding author
*NRS Medical College, Kolkata*

***Abstract***
***Background-*** *Anorectal malformation is considered to be one of the most common anomaly to be faced by paediatric surgeons. It could be easily missed and the patient present later during infancy or even childhood, more often so when the child is not suffering from severe constipation . The patients might have bowel control with or with out surgery. But, they may have problem in later life if the stool is loose or during athletic activities To provide a measure of the rate of missed anorectal malformations in the newborn period ,we have tried to review all new cases of anorectal malformation presenting to our institution after neonatal period over the past years.*
***Methods-*** *All cases of male patients newly presenting with low anorectal malformations over a 3 year span from January 2018 to January 2021 at Department of Paediatric Surgery NRS Medical College. Details about patient characteristics, presenting complaints, and treatment were noted. Patients with surgical management of the perineum done earlier were not included.*
***Results-*** *Type of low anorectal malformation was classified according to Wingspread classification. There were 3 cases of anocutaneous fistula and 6 cases of anal stenosis. 77.78% patients' mothers underwent antenatal checkup. The variety of presenting features could lead any misdiagnosis if done without the examination of perineum. Most patients presented with complaints of constipation. All the patients were managed by anoplasty except one who underwent minimal posterior saggital anorectoplasty with sigmoid colostomy.*
***Conclusion-****Good anorectal physical examination at birth can prevent misdiagnosed anorectal malformations especially low level anorectal malformation. Examination of the anal region in the newborn should involve more than just confirming the presence of an anal orifice. Attention should be taken to note the location and appearance of the anus.*
***Keywords-*** *low anorectal malformation, training traditional birth attendants ,perineum*

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Date of Submission: 09-07-2021 Date of Acceptance: 24-07-2021

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## I. Introduction

Incidence of anorectal malformation worldwide is 1 in 5000 newborns.[1] It is considered to be one of the most common anomaly to be faced by paediatric surgeons.[2] Low anorectal malformation comprises about half of all anorectal anomalies. [3] They constitute 33% patients.[4] Most of the patients with low anorectal malformations are referred to surgical care centers as newborns. Diagnosis of anorectal malformations should be established by careful perineal examination during routine neonatal examination within twenty four hours of birth. Sometimes, the diagnosis is delayed to beyond neonatal period especially in cases when the anal opening is stenotic but at or near the proper anal position.

Occasionally patients with congenital anorectal malformations have not had this condition diagnosed during the neonatal period and they presented to our department with variety of presenting complaints. A delayed diagnosis might complicate the surgical repair and may contribute to both functional and psychological problems for the patient and family. The place of delivery is a known crucial factor which affects the health and well-being of the mother and the newborn [5]. Institutional deliveries provide easy access to skilled assistance, drugs, equipment, and referral transport. India's Child Survival and Safe Motherhood Programme started in the year 1992 was concerned with training of physicians and traditional birth attendants . Traditional birth attendants have since long been assisting the women during childbirth for centuries in India. . Traditional birth attendants can contribute positively for clean delivery and clean cord care for the prevention of newborn infections, protection from loss of heat, early and exclusive breastfeeding, initiation of breathing, resuscitation, and eye care, and immunization, recognition of illness and at risk newborn referral or management and also take care of the preterm and low birth weight newborns. [6] According to World Health Organization 61% of births in India take place at home and majority of these are not attended by skilled birth attendant. [7 ]

Proper training traditional birth attendants of are required for diagnosis of anorectal malformation or else it could be easily missed and the patient present later during infancy or even childhood, more often so when the child is not suffering from severe constipation [8]. The patients might have bowel control with or with out surgery. But, they may have problem in later life if the stool is loose or during athletic activities [ 9] To provide a measure of the rate of missed anorectal malformations in the newborn period ,we have tried to review all new cases of anorectal malformation presenting to our institution after neonatal period over the past years.

## II. Methods

All cases of male patients newly presenting with low anorectal malformations over a 3 year span from January 2018 to January 2021 at Department of Paediatric Surgery NRS Medical College. A late diagnosis of low anorectal malformation was considered when a patient's disease went unrecognized after a non institutional delivery , when newborn patient was discharged from hospital without recognition of the anorectal malformation or referral to a pediatric surgeon. We included the patients that presented after one month of life. Details about patient characteristics, presenting complaints, and treatment were noted. Patients with surgical management of the perineum done earlier were not included.

## III. Results

During a span of three years nine male patients were found to present with delayed presentation of low anorectal malformation.

Careful physical examination was used to confirm the diagnosis in all the cases. The perianal region was found to be with excoriation in most of the cases. Inspection of the perineum revealed a distinctly abnormal orifice. The perineal opening was too narrow to admit an examining finger.

| | Age at diagnosis (months) | Presenting symptoms | Type of malformation | Place of birth |
|---|---|---|---|---|
| 1 | 23 | constipation | Anocutaneous fistula | Home |
| 2 | 60 | constipation | Anal stenosis | Home |
| 3 | 42 | narrow calibre stool | Anocutaneous fistula | Home |
| 4 | 10 | constipation | Anal stenosis | Home |
| 5 | 24 | constipation | Anal stenosis | Institutional |
| 6 | 9 | constipation | Anal stenosis | Nursing home |
| 7 | 2 | Abdominal distension | Anal stenosis | Institutional |
| 8 | 10 | constipation | Anal stenosis | Nursing home |
| 9 | 8 | constipation | Anocutaneous fistula | Institutional |

**TABLE 1:** CHARECTERISTICS OF THE POPULATION

Type of low anorectal malformation was classified according to Wingspread classification. There were 3 cases of anocutaneous fistula and 6 cases of anal stenosis. A stenotic anal opening may be in its normal place inside the sphincter complex, and it may be partly covered by a median bar or a membrane that is usually situated at the level of the dentate line. In males with perineal fistula the anal canal is usually situated within the external sphincter complex to a variable degree. The almost normally developed anal canal with anal crypts and transitional epithelium extends near to the anal skin. There is usually a subcutaneous fistulous communication from the anal canal to the perineal skin.

77.78% (n= 7) patients' mothers underwent antenatal checkup. The variety of presenting features could lead any misdiagnosis if done without the examination of perineum. 77.78% (n=7) patients presented with complaints of constipation. Change in posture such as partially standing position while defecating was an associated presenting complaint.42.86% (n=3) patients were firstborn to the parents.

In all the cases radial corrugations were not seen. Most of the patients (n=6) had complaints that started during weaning period of the child. All the patients irrespective of the place of delivery were not diagnosed at the time of birth.

All the patients were managed by anoplasty except one who underwent minimal posterior saggital anorectoplasty with sigmoid colostomy. In patients with a low malformation, a primary repair is usually possible. A diverting colostomy and staged surgery is reserved for patients who are clinically unstable, if there is no expertise available to perform primary surgery or if the fistula is stenotic and difficult to dilate prior to anoplasty.

## IV. Discussion

During this period of three years nine male patients were found . Consistent findings amongst the members of the study were observed in terms of clinical presentation and age at diagnosis. It was found that most of the patients suffered from chronic constipation that became worse or refractory to medical treatment

around the time of dietary change from breast milk to formula or at the time of weaning or introduction of semisolid food .This could be due to the fact that there is greater difficulty of passing the more solid stool rather than liquid through the abnormal anal opening. Earlier observation is important because it has not been reported previously in a group of patients; indeed, it has been stated that conservative treatment of anterior ectopic anus is generally adequate.[10]

Many a times it is difficult to distinguish a normal from an abnormal anus. Usually the abnormal anus is located more anteriorly and sometimes lateral to the midline. There is absence of radial corrugations produced by the underlying anal sphincter. However, in the older infant or child, it is important to correlate the appearance of the anus with its function .[3]

Diagnosis of anorectal malformation during antenatal is very rare. It is below 15% of the cases [11]. On ultrasonographic examination, direct findings include no perianal muscular complex and no target sign that is presence of hypoechoic anal sphincter and echogenic anal mucosa. Indirect findings include dilated distal bowel segments and calcified intraluminal meconium in second and third trimester [12]. Three dimensional ultrasonography is more accurate to determine the type of anorectal malformation. 3 dimensional reconstruction is used to specify the location of an ectopic anal sphincter. Fetal Magnetic resonance imaging is considered as an excellent supplementary examination technique to confirm the presence of bowel dilatation [ 13 ]. During palpation, the size of anus is noted and its location within anal sphincter complex then con firm it by cutaneous anal reflex or electromyography if in doubt. Rigid anal is abnormal, which normal anus will stretch after a slow and gentle insertion with a well lubricated little finger. Meconium that went out in less than 24 hours of age should be noted [14, 15] .

Minimal posterior saggital anorectoplasty and minimal posterior sagittal anoplasty is the current standard technique for the management of the condition. [ 16]. Most of the patients we encountered were managed by anoplasty. They were managed by regular anal dilatation. There was no mortality in the study group . Delayed diagnosis of low anorectal malformation has the ability to contribute to unnecessary patient suffering and to a certain extent decreased parental confidence in their physician.It is an important fact that not all anorectal malformations will present in the newborn period as an imperforate anus. Examination of the anal region in the newborn should involve more than just confirming the presence of an anal orifice. Attention should be taken to note the location and appearance of the anus.

## V. Conclusion

Developed countries have occasional reports of delayed presentation of such anorectal malformation. The situation in the developing countries like India is different where a significant numbers of deliveries take place at home assisted by nonmedical or trained birth attendants . Good anorectal physical examination at birth can prevent misdiagnosed anorectal malformations especially low level anorectal malformation. Examination of the anal region in the newborn should involve more than just confirming the presence of an anal orifice. Attention should be taken to note the location and appearance of the anus.

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