Original Research Article

# Clinical presentation of anorectal malformations at a tertiary care centre in Marathwada region: An observational study

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## Abstract

**Background:** The anorectal malformations (ARM) are a variety of congenital defects involving a structural defect in anus, rectum and variable segments of the urogenital system. The present study describes the clinical presentation of the anorectal malformations at the tertiary care centre in Marathwada region. **Methods:** This prospective observational study was conducted at tertiary care hospital between 2013 and 2015 in department of general surgery. Babies of anorectal malformations admitted and treated at our centre were included in the study. The diagnosis was made on the basis of clinical examination, and/or invertogram, abdominal and perineal USG. A nasogastric tube was used to exclude oesophageal atresia and a piece of gauze was placed on the tip of the penis to check for presence of meconium particles and if not present, urine examination was done. **Results and Conclusions:** In present study, 57% were males and 43% were females and sex ratio was 1.23:1 showing male sex predisposition. Maximum cases (73%) presented in the early neonatal period. In the present study, 90% babies were full term and 10% babies were preterm deliveries. Also, 10% mothers had oligohydramnios, 3% mothers had polyhydramnios, and 87% mothers had no significant antenatal history. Among the study subjects, 91% cases had no family history with anorectal malformations, 7% cases had 1st sibling and 2% cases had 2nd sibling with anorectal malformations. In the present study, 49% cases presented with absent anus with abdominal distension, 37% of cases presented with complaints of stool coming from vagina and 14% cases had presentation of ectopic anus. Genitourinary anomalies were the most common associated anomalies (28%) found associated with the anorectal malformations. The present study provides an insight regarding the clinical profile of the anorectal malformation cases from our tertiary care centre in Marathwada region of Maharashtra

**Keywords:** Vestibular anus, Invertogram, Ectopic anus.

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## INTRODUCTION

The anorectal malformations (ARM) are a variety of congenital defects involving a structural defect in anus, rectum and variable segments of the urogenital system. These are among the most frequent congenital structural malformations across the world managed by the paediatric surgeons. The diagnosis of the ARM before birth is rare and most of the cases are diagnosed in the early neonatal period. The clinical features and associated malformations are important factors in the prognosis and outcome of the surgical management.[1-3] There is a need for data regarding the clinical profile of anorectal malformations from various centres and different regions of the country for better understanding the problem and help in designing mitigation strategies. The present study describes the clinical presentation of the anorectal malformations at the tertiary care centre in Marathwada region.

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## METHODS

This prospective observational study was conducted at tertiary care hospital between 2013 and 2015 in department of general surgery. Babies of anorectal malformations admitted and treated at our centre were included in the study. The diagnosis was made on the basis of clinical examination, invertogram, and/or abdominal and perineal USG. A nasogastric tube was used to exclude oesophageal atresia and a piece of gauze was placed on the tip of the penis to check for presence of meconium particles and if not present, urine examination was done.

## RESULTS

In present study, 57% were males and 43% were females and sex ratio was 1.23:1 showing male sex predisposition. Table 1 shows the age at presentation of the study subjects. In present study, 90% babies were full term and 10% babies were preterm. Among the study subjects, 4% babies were <2kg, 57% babies were between 2-3 kg, and there were 39% cases weighing between 3-4 kg. Range was from 1.7- 4kg and average was 2.77kg. In the present study, 10% mothers had oligohydramnios, 3% mothers had polyhydramnios, and 87% mothers had no significant antenatal history. Among the study subjects, 91% cases had no family history with anorectal malformations, 7% cases had 1st sibling and 2% cases had 2nd sibling with anorectal malformations. In the present study, 49% cases presented with complaints of absent anus with abdominal distension, 37% of cases presented with complaints of stool coming from vagina (vestibular anus) and 14% cases had presentation of ectopic anus (abnormal location of anus excluding vestibular anus). In 15% cases of anorectal malformations in the study, colostomy was already done (11% cases-absent anus and 4% cases-vestibular anus). Table 2 shows the associated anomalies among the anorectal malformation cases.

**Table 1:** Age at presentation of the study subjects

| Sr. No. | Age distribution | Percentage |
|---|---|---|
| 1 | Early neonatal | 73% |
| 2 | Late neonatal | 1% |
| 3 | Post neonatal | 22% |
| 4 | After infancy | 4% |

**Table 2:** The associated anomalies in Anorectal malformation subjects

| Sr. no. | Associated anomalies | Percentage |
|---|---|---|
| 1 | Vertebral | 7% |
| 2 | Cardiac | 4% |
| 3 | Genitourinary | 28% |
| 4 | Tracheo-esophageal | 1% |
| 5 | Limb and other | 8% |
| 6 | No anomalies | 52% |

## DISCUSSION

In the present study, male sex predisposition was observed with a sex ratio of 1.23:1 (57% males, 43% females). In a recent study by Shenoy NS *et al.* from Kolkata, in eastern India, 63% were found to be males and 37% were females. Similarly, from other parts of the world also, it has been reported that there is male preponderance in anorectal malformations ranging from 55 to 71%.[1,5-7] In our study, maximum cases (73%) presented in the early neonatal period. So, there were 27% babies who presented beyond early neonatal period and they were grouped in as delayed presenters. Sinha SK *et al.* study from 2003 to 2006 at Lucknow, India reported that 25% cases of anorectal malformations were delayed presenters.[8] They mentioned that delayed diagnosis can adversely impact the management. They highlighted that wrong advice regarding correct age of treatment is a frequent cause of delayed presentation. Developed countries have only occasional cases of late presentation. Thus, there is need for awareness regarding anorectal malformations and its treatment protocol among the public and health caregivers. Lawal T *et al.* review of African studies also found that delayed presentation is common in African countries. They have also mentioned that delayed presentation has an adverse impact on the prognosis and management.[1] In our study, Genitourinary anomalies were the most common associated anomalies (28%) found associated with the anorectal malformations. Bhargava P *et al.* study (1999 and 2000) from New Delhi also reported that urogenital anomalies were the most common associated anomalies and they observed it in 24% cases.[9] Almaramhy HH study (2011) from Saudi Arabia also reported that genitourinary anomalies were commonly associated with anorectal malformation. They observed that 34% cases were having associated genitourinary anomalies.[7] Mirza B *et al.* study (2008-09) from Lahore, Pakistan reported that there was male preponderance (3.35:1) in patients with anorectal malformations and urogenital anomalies were the commonest anomalies associated with it.[10]

Limitations of the present study are a hospital based small sample and cross-sectional design. Further more elaborate and robust studies are needed on the subject. The present study however provides an insight regarding the clinical profile of the anorectal malformation cases from our tertiary care centre in Marathwada region of Maharashtra.

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