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Research Article

A SINGLE CENTRE STUDY OF ANOMALIES ASSOCIATED WITH ANORECTAL MALFORMATIONS

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A B S T R A C T

Background: Anorectal malformations (ARM) are frequently accompanied by other congenital anomalies**.** The aimof the study was to compare occurrence of associated anomalies in patients with anorectal malformations and classify the subjects according to Krickenbeck's classification of anorectal malformations.

Methods: An observational retrospective study in 2020-21 was conducted in the Department of Pediatric Surgery at LokmanyaTilak Municipal Medical college and Municipal Hospital, Sion, India. The study was approved by the institutional ethics committee. 90 patients including newborns upto the age of 12 years diagnosed with anorectal malformations were considered in the study. Patients were stratified according to the Kricken beck's classification of anorectal malformations. The associated anomalies with anorectal malformations were cardiovascular, genital, urinary, respiratory, gastrointestinal, central nervous system and skeletal.

Results: We assessed 90 patients; 59% patients were males. 77% patients with anorectal malformation had at least one other associated anomaly. The most common types of anorectal malformations in the present study, according to Krickenbeck's classification were rectovestibular and rectovesical fistulae. The urinary system anomalies such as vesicoureteric reflux and renal agenesis followed by cardiovascular system anomalies such as atrial septal defect were the most common anomalies associated with anorectal malformations. 43% patients had urinary system anomalies; 38% had cardiovascular anomalies; 10% had gastrointestinal system anomalies; 12% had genital system anomalies; 24 % had central nervous system anomalies and 4% patients had skeletal anomalies associated with anorectal malformations respectively.10% patients with anorectal malformations had associated VACTERL syndrome.

Conclusion: The incidences of many associated anomalies with anorectal alformations in our study were higher than those compared with the earlier studies. A detailed physical, systemic and radiological examination along with multidisciplinaryapproach is required in all patients withanorectal malformation. Genetic counselling may be necessary in complex cases

INTRODUCTION

Anorectal malformations (ARM) is a common congenital anomalies encountered by pediatric surgeons, with incidence ranging between 1 in 2000 to 1 in 5000 live births. (1–3)

In 2005, a new international standardized diagnostic classification was devised by the Krickenbeck Conference on ARM.(4) This system incorporates an anatomic description of the ARM.

Anorectal malformation is associated with wide spectrum of other congenital abnormalities involving genitourinary, spinal, cardiovascular, gastrointestinal, craniofacial, skeletal and other systems.(3,5–7)

Association of other malformations in patients with anorectal malformation vary between 20% to 70% according to the literature.(8,9)

The associated anomalies can be serious and in some instances, can bear serious implications on the long-term prognosis of patient in comparison to the anorectal malformation itself. Therefore, early detection of such anomalies and timely intervention is important to improve outcome.(10) The present study highlights various associated anomalies in patients with anorectal malformations in a geographically defined population.

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Aim: To study frequency of associated anomalies in patients with anorectal malformations

Objectives

1. To classify patients with anorectal malformations according to Krikenbeck's classification
2. To study associated anomalies by organ system in patients with anorectal malformations.

MATERIALS & METHODS

The present study was retrospective observational study conducted at LTMMC and LTMGH, Sion, Mumbai. Pediatric patients between newborn to 12 years diagnosed with anorectal malformation were included in the study. This study was started after ethical committee clearance. All required data was collected from submitted case records of anorectal malformation patients. Patients were classified according to Kricken beck's classification for anorectal malformation. Those with rectobulbar and rectoprostaticfistul as were considered in the same group of rectourethralfistulas. Cardiovascular, genital, urinary, respiratory, gastrointestinal, central nervous system and skeletal anomalies associated with anorectal malformations were studied.

RESULTS

A total of 90 patients were included in this study. There were 53 (59%) male patients and 37 (61%) female patients.

Overall, 69 patients had at least one associated malformation leaving only 21 patients (23%) with isolated ARM.

Most common type of anorectal malformation according to Kricken beck's anatomic classification in our study was rectovestibular fistulae -23 (26%), followed by rectovesical fistulae – 20 (22%) patients, rectourethralfistulas - 19 (21%) patients, perinealfistula– 11 (12%) patients.9 (10%) patients had no fistulae. There were 8(9%) female patients who had persistent cloacalanomaly.

Table no 1 Patients of ARM according to Kricken beck's anatomic classification

Type of ARM according to Krickenbeck's classification	Number (n=90)	Male (n=53)	Female(n=37)
Perineal fistulae	11	5	6
Rectovesical fistulae	20	20	0
Rectourethral fistulae	19	19	0
Recto-vestibular fistulae	23	NA	23
No fistulae	9	9	0
Persistent cloaca	8	NA	8

Urinary, gastrointestinal, central nervous system and respiratory malformation were most commonly associated with rectovesical fistulae. Genital malformations were commonly associated with persistent cloaca and perineal fistulae. Cardiac malformation was most common finding in rectourethral fistulae group. Skeletal malformation was common in no fistulae group.

Urinary system anomalies

Total 39 patients had one or more urinary tract anomalies and urinary system was most commonly affected system. Most common encountered anomaly was vesicoureteric reflux (13 patients) followed by renal agenesis (10 patients). (Fig.-A)

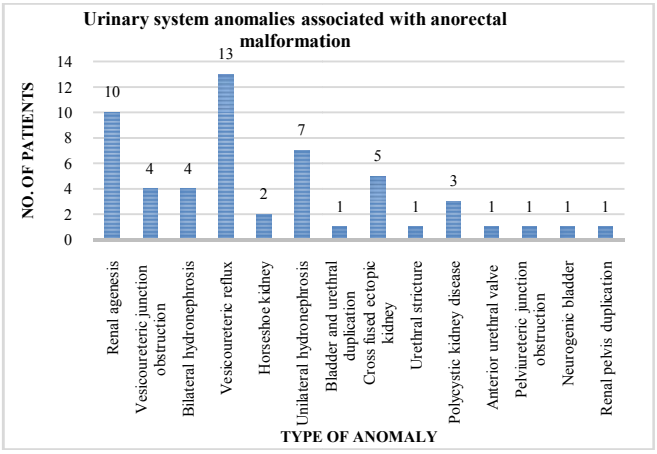

Fig (A)

Cardiovascular system anomalies

Second most common associated anomaly in our study was cardiac anomaly. Total 34 patients were affected with single or multiple anomalies with atrial septal defect being most common (30 patients).(Fig.-B)

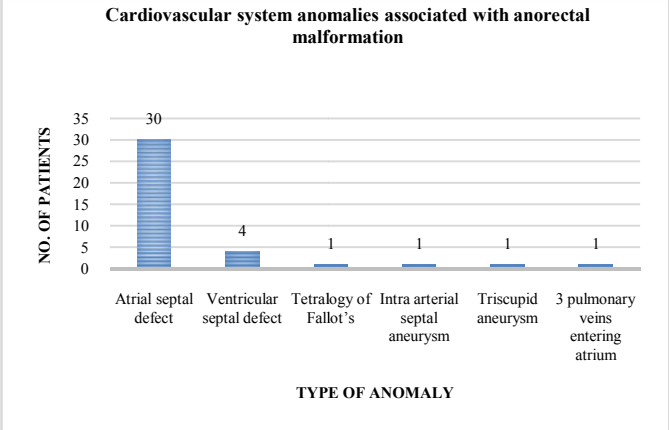

Fig (B)

Table no 2 Number of malformations seen in association with anatomic type of ARM according to the Krickenbeck classification.

Type of ARM	Urinary Malformations	Genital malformations	Cardiac malformations	Gastrointestinal malformations	Skeletal malformations	Central nervous system malformations	Respiratory system malformation
Perineal fistulae	3	3	5	0	1	3	0
Rectourethral fistulae	9	2	8	2	1	4	0
No fistulae	5	1	4	1	2	2	0
Persistent cloaca	6	3	3	2	0	2	0
Rectovestibular fistulae	6	0	7	1	0	4	0
Rectovesical fistulae	10	2	7	3	0	7	1

Gastrointestinal system anomalies

Gastrointestinal anomalies were seen in 9 patients, most common anomaly was pouch colon (Fig.- C)

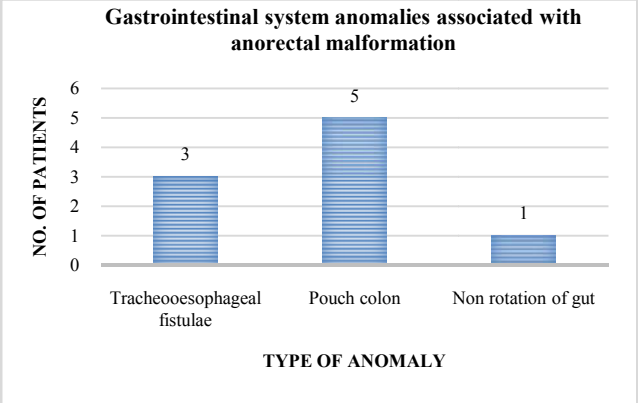

Fig (C)

Genital system anomalies

11 patients had one or more genital anomalies. Amongst which hypospadias was most common seen in 5 patients. (Fig.-D)

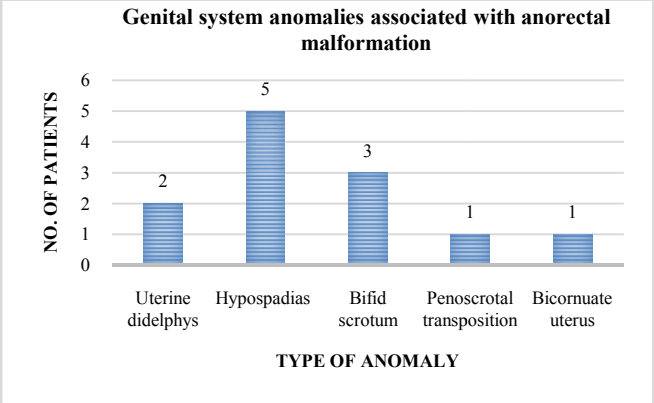

Fig (D)

Central nervous system anomalies

22 patients had one or more central nervous system anomalies, 4 patients had of spina bifida. Incomplete sacrum (6 patients) and hemivertebrae (5 patients) was most common vertebral anomaly found in our study.(Fig.-E)

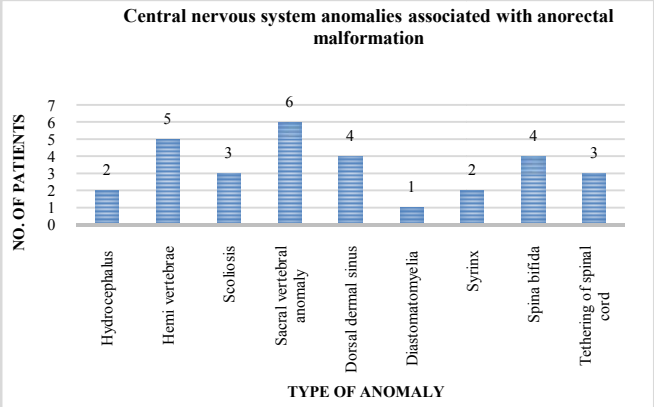

Fig (E)

Skeletal anomalies

4 patients had one or more associated skeletal anomalies. (Fig.-F)

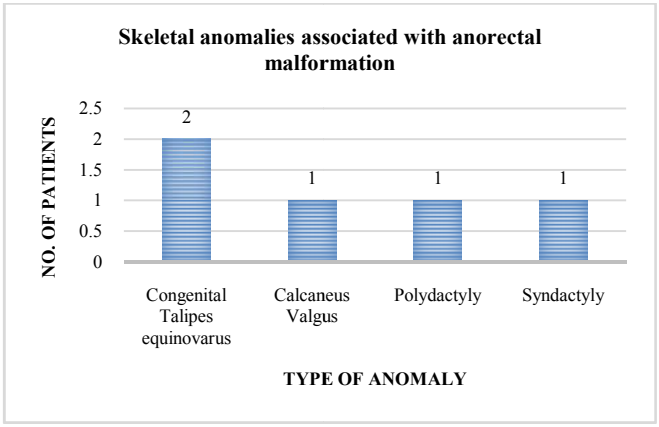

Fig (F)

VACTERL syndrome

Table 3 VACTERL syndrome associated with anorectal malformations

Type of ARM	Number of patients
High	3
Intermediate	None
Low	6

VACTREL association was seen in 10% patients, with more common association in low ARM group in our study.

DISCUSSION

In relation to the anatomic form of ARM as described by the Krickenbeck classification, our analysis demonstrates the occurrence of associated abnormalities seen in ARM in a subset of 90 patients seen at a tertiary care hospital in west coast of India.

In our study, the proportion of male patients with ARM was higher (59%) which was comparable to the other studies. Boys commonly suffered from rectovesical and rectourethral fistulae. Most common anomaly in female patients was rectovestibular fistulae.

Only 21 patients (23%) were suffering from isolated ARM. Anorectal malformation with associated other abnormalities was more common (77%) compared to isolated ARM in our research which is comparable with various studies carried out over years where the percentage of patients with associated anomalies ranged from 30-71%. (11)

In study done by Shenoy *et al*, 56.04% of the patients had associated other system anomalies along with ARM. (12)Nah *et al* in his study found 78% of children with ananorectalmalformation had other associated anomalies. (13)

Urinary abnormalities

The most common organ system that was affected by additional anomalies was the urinary system affecting 43%ofour patients. Urinary anomaly was most frequent anomaly in study done by Ratan *et al* affecting 31% patients. (5)

Different studies in literature found that frequency of genitourinaryanomalies ranges anywhere from 26% to 60% in patients with ARM.(6–8,14–16)

The most frequent genitourinary abnormality found in our patients was VUR in 13 patients. Similar observation was found in study done by Nah *et al*.(13)

Urinary malformations were seen in all groups but highest with rectovesical fistulae followed by recto urethral fistulae group. Different studies have concluded that there is decreasing incidence of genitourinaryabnormalities with diminishing complexity of the ARM, which is consistent in our study.

In other studies incidence of genitourinary anomalies was highest in the rectovesicalfistula group.(13,17)

Cardiovascularanomalies

We observed cardiovascularanomaliesin38%ofthepatientswithARM. Cardiovascularanomalieswereobservedin 28.3%ofthepatientswithARM in study done by stoll *et al*.(3)Thereportedrange inotherseriesvariesinbetween6and 27%. (7,8,14–16)

We found that most common cardiovascular anomaly was atrial septal defect followed by ventricular septal defect. Cardiac anomalies were commoner with rectovesical fistulae, rectourethral fistulae and rectovestibular fistulae in our study. Nah *et al* found atrial septal defect was most common associated cardiac anomaly with rectourethral and rectovestibular fistulae groups being more commoly affected.(13)

Central nervoussystemabnormalities

Inourinstitution, ultrasonography of the spine, skull and Xray spine is carried outinallinfants as screening tool. MRI of the spine is carried out only in required cases asgeneralanesthesia is required. As there is a known association of spinaldysraphism with ARM, screening should be done during initial assessment so that neurosurgical correction can be planned although its effect on bowel function is still unclear.

Common found vertebral anomalies were incomplete sacrum and hemi vertebrae. In our study,17% patients had vertebral anomalies on radiographs compared to 28% patients in study of Ratan *et al*. (5)

Spina bifida was seen in 4 cases (one from each group-recto vestibular, rectoperineal, no fistulae and cloaca). Two patients suffered from hydrocephalus. In our study the higher number of CNS anomalies was found with rectovesical fistulae patients. Nah *et al* in his study concluded that CNS anomalies were common with cloaca followed by recto vestibular fistula and rectovesical fistulae**.**(13)

Musculoskeletal anomalies

Musculoskeletal anomalies were observed in 4% patients in our series. There- ported range of musculoskeletal anomalies in patients with ARM is15to44%(7,8,14,15) and12.3% in the series of Cuschieri *et al*.(18)

Gastrointestinal anomalies

Gastrointestinal anomalies were uncommon with pouch colon (5.5%) being most common gastrointestinal anomaly.

Association of tracheoesophageal fistula, in our study was 3.33% whereas in various series observed range is between 5.2- 9.59%.(19–21)

Genital anomalies

Most common genital anomaly in our study group was hypospadias followed by bifid scrotum. Genital abnormalities were commoner with persistent cloaca and rectoperineal fistulae group.

Goossens et at found that the highestincidence of genital anomalies was seen in rectovesicalfistulagroup.(17)

Respiratory system anomaly was the least common associated anomaly in our study. Only one patient was affected and had subglottic tracheal stenosis.

Frequently associated other organ systems anomalies with ARM is referred to VACTERL association (Vertebral; Anorectal; Cardiac; Tracheo-Esophageal fistula; Renal; Limb). Reported incidence of such association ranges from 30% to 60% for low lesions (7,22,23) and approximately 70% for high lesions.(7)However, reported incidence in patients with ARMs with VACTERL syndrome (3 or more anomalies) ranges from 5% to 31%.(3,8,13,22)

VACTERL syndrome association in our study was found in 10% patients.Totonelli *et al* reported VACTERL association in17.8 %(24), similar to what has been reported by Cuschieri *et al*. in their large sample population.(18)

CONCLUSION

In our study population, we demonstrated that77%ofchildren with ananorectalmal formation have other associated anomalies, which is higher as compared to other reports. More than one organ system is involved in many patients. VACTREL association was seen in 10% patients.

Urinary system is most common affected organ system with anorectal malformation with more frequent involvement with rectovesical and rectourethral fistula group. Other common affected organ system were cardiac and central nervous system Associated malformation was highest in those with rectovesical fistulae patients followed by rectourethral fistula group.

A detailed physical, systemic and radiological examination along with multidisciplinaryapproach is required in allpatients withanorectal malformation in neonatal period itself to reduce future morbidity and mortality regardless of type of ARM with particular focus on urinary, cardiovascular and central nervous system abnormalities.

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