INTERNATIONAL JOURNAL OF CURRENT ADVANCED RESEARCH
ISSN: O: 2319-6475, ISSN: P: 2319-6505, Impact Factor: 6.614
Available Online at www.journalijcar.org
Volume 11; Issue 09(x); xxxx2022; Page No. 1551-1555
DOI: http://dx.doi.org/10.24327/ijcar.2022.1555.0347

Research Article

A SINGLE CENTRE STUDY OF ANOMALIES ASSOCIATED WITH ANORECTAL MALFORMATIONS

Akshay Baheti, Shahaji Deshmukh, Apoorva Kulkarni, Abhaya Gupta, Sanjay Oak and Paras Kothari

Department of Pediatric Surgery, LTMMC and LTMGH Hospital, Sion, Mumbai

A R T I C L E I N F O

Article History:

Received 15th August, 2022
Received in revised form 25th August, 2022
Accepted 20th September, 2022
Published online 28th September, 2022

Keywords:

A B S T R A C T

Background: Anorectal malformations (ARM) are frequently accompanied by other congenital anomalies**.** The aim of 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 Lokmanya Tilak 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 Krickenbeck'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 multidisciplinary approach is required in all patients with anorectal 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.

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 recto bulbar and rectoprostatic fistulas were considered in the same group of rectourethralfistulas.

Corresponding author:* **Akshay Baheti
Department of Pediatric Surgery, LTMMC and LTMGH Hospital, Sion, Mumbai

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 recto vestibular fistulae -23 (26%), followed by rectovesical fistulae -20 (22%) patients, recto urethral fistulas - 19 (21%) patients, perineal fistula-11 (12%) patients.9 (10%) patients had no fistulae. There were 8 (9%) female patients who had persistent cloacal anomaly.

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)

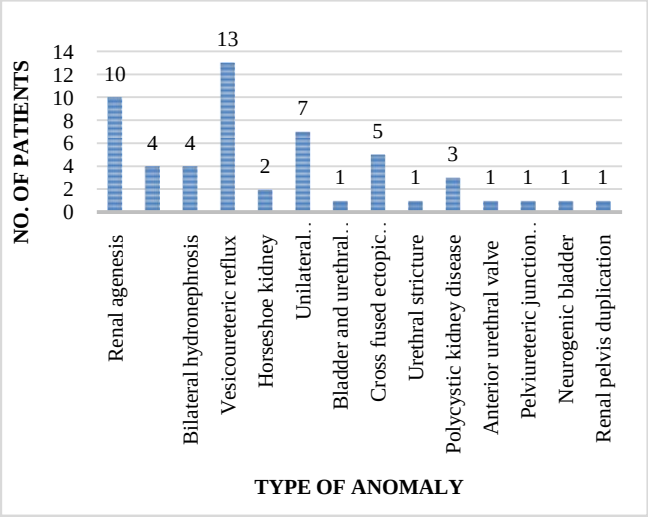

Fig A Urinary system anomalies associated with anorectal malformation

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)

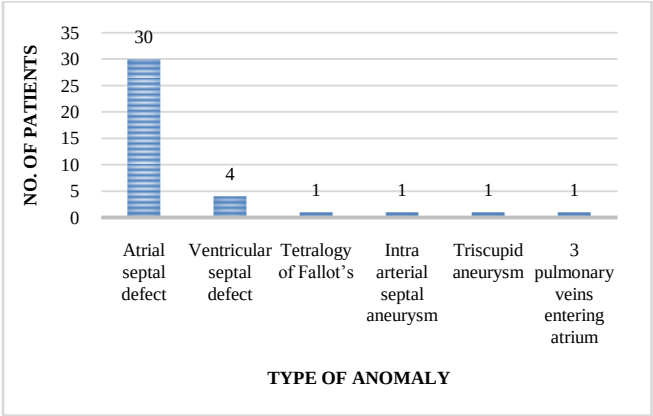

Fig B Cardiovascular system anomalies associated with anorectal malformation

Gastrointestinal system anomalies

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

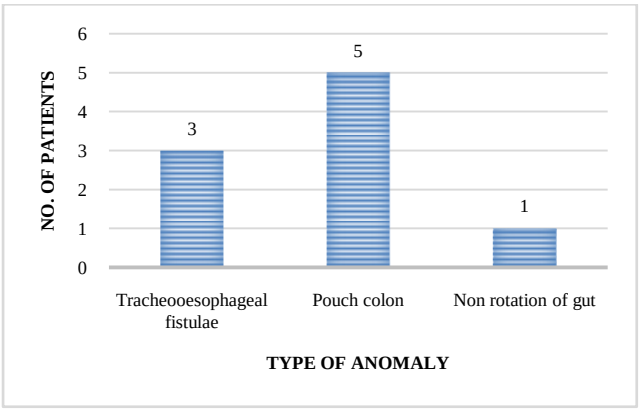

Fig C Gastrointestinal system anomalies associated with anorectal malformation

Genital system anomalies

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

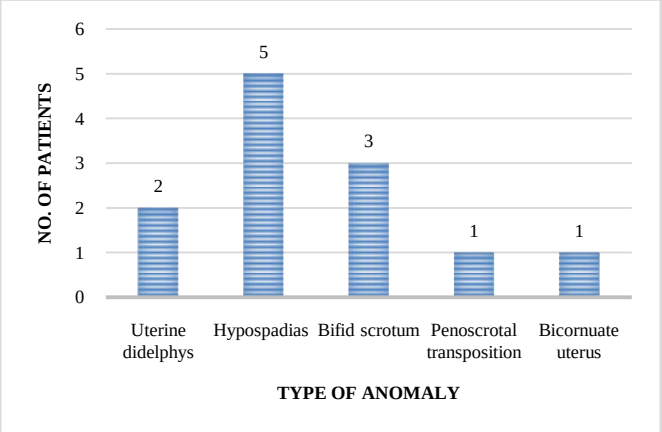

Fig D Genital system anomalies associated with anorectal malformation

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)

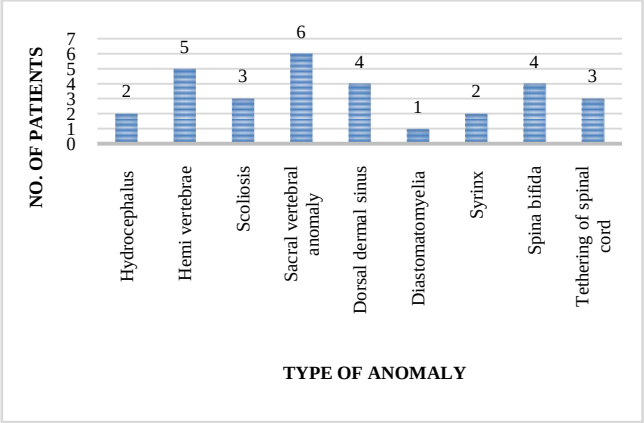

Fig E Central nervous system anomalies associated with anorectal malformation

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]

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

Skeletal anomalies

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

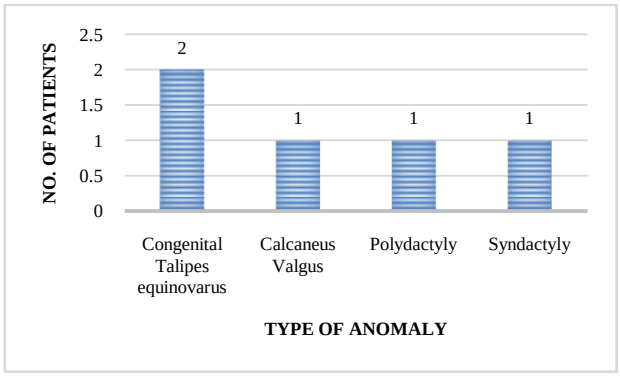

Fig F Skeletal anomalies associated with anorectal malformation

VACTERL syndrome

Table 3 VACTERL syndrome associated with anorectal malformations

Type of ARM	Number of patients
High	3

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 an anorectal malformation 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 genitourinary anomalies 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 rectourethral fistulae group. Different studies have concluded that there is decreasing incidence of genitourinary abnormalities 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]

Cardiovascular anomalies

We observed cardiovascular anomalies in 38% of the patients with ARM. Cardiovascular anomalies were observed in 28.3% of the patients with ARM in study done by stoll *et al*.(3)The reported range in other series varies in between 6 and 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 atrialseptal defect was most common associated cardiac anomaly with rectourethral and rectovestibular fistulae groups being more commonly affected.[13]

Central nervous system abnormalities

In our institution, ultrasonography of the spine, skull and X ray spine is carried out in all infants as screening tool. MRI of the spine is carried out only in required cases as general anesthesia is required. As there is a known association of spinal dysraphism 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% patientsinourseries. There- ported range of musculoskeletal anomalies in patients with AR Mis 15 to 44% [7,8,14,15] and 12.3% 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 highest incidence of genital anomalies was seen in rectovesicalfistula group.[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 that 77% of children with an anorectal malformation have other associated anomaly, which is higher as compared toot her 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 recto urethral fistula group.

A detailed physical, systemic and radiological examination along with multidisciplinary approach is required in all patients with anorectal 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.

References

1. Levitt MA, Peña A. chapter 36 - IMPERFORATE ANUS AND CLOACAL MALFORMATIONS. In: Holcomb GW, Murphy JP, Ostlie DJ, editors. Ashcraft's Pediatric Surgery (Fifth Edition) [Internet]. Philadelphia: W.B. Saunders; 2010 [cited 2022 Feb 13]. p. 468–90. Available from: https: //www. sciencedirect.com/science/article/pii/B9781416061274000367
2. Jenetzky E. Prevalence estimation of anorectal malformations using German diagnosis related groups system. Pediatr Surg Int. 2007 Dec 1;23(12):1161–5.
3. Stoll C, Alembik Y, Dott B, Roth MP. Associated malformations in patients with anorectal anomalies. European Journal of Medical Genetics. 2007 Jul 1;50(4):281–90.
4. Holschneider A, Hutson J, Peña A, Beket E, Chatterjee S, Coran A, *et al*. Preliminary report on the International Conference for the Development of Standards for the Treatment of Anorectal Malformations. Journal of Pediatric Surgery. 2005 Oct 1;40(10):1521–6.
5. Ratan SK, Rattan KN, Pandey RM, Mittal A, Magu S, Sodhi PK. Associated congenital anomalies in patients with anorectal malformations—a need for

developing a uniform practical approach. Journal of Pediatric Surgery. 2004 Nov 1;39(11):1706–11.

6. Joseph VT, Chan KY, Siew HF. Anorectal malformations and their associated anomalies. Annals of the Academy of Medicine, Singapore. 1985;14(4):622–5.
7. Endo M, Hayashi A, Ishihara M, Maie M, Nagasaki A, Nishi T, *et al*. Analysis of 1,992 patients with anorectal malformations over the past two decades in Japan. Journal of Pediatric Surgery. 1999 Mar 1;34(3):435–41.
8. Mittal A, Airon RK, Magu S, Rattan KN, Ratan SK. Associated anomalies with anorectal malformation (ARM). Indian J Pediatr. 2004 Jun 1;71(6):509–14.
9. Spouge D, Baird PA, Opitz JM, Reynolds JF. Imperforate anus in 700,000 consecutive liveborn infants. American Journal of Medical Genetics. 1986;25(S2):151–61.
10. Nievelstein RAJ, Vos A, Valk J. MR imaging of anorectal malformations and associated anomalies. Eur Radiol. 1998 May 1;8(4):573–81.
11. Byun SY, Lim RK, Park KH, Cho YH, Kim HY. Anorectal Malformations Associated with Esophageal Atresia in Neonates. Pediatr Gastroenterol Hepatol Nutr. 2013 Mar 31;16(1):28–33.
12. Shenoy NS, Kumbhar V, Basu KS, Biswas SK, Shenoy Y, Sharma CT. Associated anomalies with anorectal malformations in the Eastern Indian population. Journal of Pediatric and Neonatal Individualized Medicine (JPNIM). 2019;8(2):e080214–e080214.
13. Nah SA, Ong CC, Lakshmi NK, Yap T-L, Jacobsen AS, Low Y. Anomalies associated with anorectal malformations according to the Krickenbeck anatomic classification. Journal of pediatric surgery. 2012;47(12):2273–8.
14. Boocock GR, Donnai D. Anorectal malformation: familial aspects and associated anomalies. Archives of Disease in Childhood. 1987 Jun 1;62(6):576–9.
15. Cho S, Moore SP, Fangman T. One hundred three consecutive patients with anorectal malformations and their associated anomalies. Archives of pediatrics & adolescent medicine. 2001;155(5):587–91.
16. Hassink EAM, Rieu PNMA, Hamel BCJ, Severijnen RSVM, Staak FHJ vd, Festen C. Additional congenital defects in anorectal malformations. Eur J Pediatr. 1996 Jun 1;155(6):477–82.
17. Goossens WJH, de Blaauw I, Wijnen MH, de Gier RPE, Kortmann B, Feitz WFJ. Urological anomalies in anorectal malformations in The Netherlands: effects of screening all patients on long-term outcome. Pediatric surgery international. 2011;27(10):1091–7.
18. Cuschieri A, Group EW. Anorectal anomalies associated with or as part of other anomalies. American journal of medical genetics. 2002;110(2):122–30.
19. Khoury MJ, Cordero JF, Greenberg F, James LM, Erickson JD. A population study of the VACTERL association: evidence for its etiologic heterogeneity. Pediatrics. 1983;71(5):815–20.
20. Kiesewetter WB, Turner CR, Sieber WK. Imperforate anus: review of a sixteen year experience with 146 patients. The American Journal of Surgery. 1964;107(3):412–21.
21. Quan L. The VATER association: vertebral defects, anal atresia, tracheoesophageal fistula with esophageal atresia, radial dysplasia. Birth Defects. 1972;8:75–8.
22. Javid PJ, Barnhart DC, Hirschl RB, Coran AG, Harmon CM. Immediate and long-term results of surgical management of low imperforate anus in girls. Journal of pediatric surgery. 1998;33(2):198–203.
23. Pakarinen MP, Rintala RJ. Management and outcome of low anorectal malformations. Pediatric surgery international. 2010;26(11):1057–63.
24. Totonelli G, Catania VD, Morini F, Fusaro F, Mosiello G, Iacobelli BD, *et al*. VACTERL association in anorectal malformation: effect on the outcome. Pediatric surgery international. 2015;31(9):805–8.

How to cite this article:

Akshay Baheti ***et al*** (2022) ' A Single Centre Study of Anomalies Associated with Anorectal Malformations', *International Journal of Current Advanced Research*, 11(09), pp. 1551-1555. DOI: http://dx.doi.org/10.24327/ijcar.2022.1555.0347

******