Original research article

A study on association between abnormalities of urogenital tract with ano-rectal malformations (arm's) in a tertiary care centre

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Abstract

Background: Ano-rectal malformations (ARM) are congenital development disorders commonly associated with anomalies of other systems such as VACTERL syndrome. This study aims to evaluate the association between ARM's and uro-genital abnormalities in neonates presenting to our tertiary care center.

Materials and methods: 100 neonates with ano-rectal malformations were included in this prospective observational study which was conducted over 1 year period by the Department of Pediatric surgery, Niloufer Hospital, Hyderabad.

Results: most common type of ARM was high type. Males constituted the majority of the study population (65%). Urological abnormalities were more common than genital abnormalities. Most common urological abnormality was vesico-ureteric reflex, followed by hydronephrosis. Most common genital abnormality observed in present study was cryptorchidism in males and bicornuate uterus in females.

Conclusion: The study emphasizes on screening for uro-genital abnormalities in patients with ARM's in lieu of its high incidence and degree of association, especially in males.

Keywords: Ano-rectal anomalies, uro-genital anomalies, vesico-ureteric reflex, cryptorchidism, uterus

Introduction

Patients with anorectal anomalies usually have an imperforate anus with the anal canal opening as a fistulous tract opening either anterior to the usual anal opening or into any of the adjacent structures (such as urinary tract in males or gynecological structures in females). The incidence of ano-rectal malformations is 1 for every 1000 live births with a higher male preponderance (1.2:1). The most common ano-rectal malformation is recto-urethral fistula in males and recto-vestibular fistula in females ^[1]. Management of imperforate anus should be done by identifying the type of ARM, look for any other abnormality and then correcting them simultaneously ^[2, 3]. Although there are many studies reporting urogenital anomalies in patients with ARM, this study aims to evaluate the types of ARM and their association with urogenital abnormalities.

Methodology

This prospective study was conducted in the department of pediatric surgery, Niloufer hospital Hyderabad, over 1 year, i.e. from April 2022 to March 2023. All neonates with anorectal-malformations presenting during the study period were included in the study.

Patients whose parents did not give consent to participate in the study were excluded from the study.

A detailed history was taken with special emphasis on family history, birth history and antenatal history. General examination and systemic examination was done for all the neonates to identify any other malformations or syndromes. The type of urogenital anomalies were noted.

All neonates were subjected to radiographic evaluation to assess the level of ano-rectal anomalies-high, intermediate or low levels. Intra-venous pyelography (IVP) and voiding cystourethrography (VCUG) were done in patients with intermediate and high levels of ano-rectal malformations and in patients with low level of ano-rectal malformations, urogenital anomalies were defined as that of penis, testis or scotal areas in males and uterus, cervix and vagina in females.

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A total of 100 patients with ano-rectal malformations were included in the study. Ethical committee approval was taken prior to start of the study. A written informed consent was taken from the parents of the all the neonates. Data was analyzed using Microsoft excel.

Results

100 neonates with ano-rectal malformations were included in this study. Males constituted the majority of the study group (65%) and females constituted the remaining 35%. Among the 100 patients, 40% had high level anomalies, 35% had intermediate level and 25% had low level of ano-rectal anomalies.



Fig 1: Distribution of type of anorectal anomalies

All 75 patients with high and intermediate ano-rectal anomalies were subjected to renal ultrasound examination and voiding cysto-urethrography (VCUG). The remaining 25 patients with low anorectal anomalies were examined for genital tract abnormalities.

Renal ultrasonography detected urological anomalies in 35 patients, while VCUG detected 45 patients with urological anomalies. Thus a total of 45 patients had urological anomalies of the 75 patients (45%). Genital anomalies were observed in 20 patients with low anorectal malformations (20%).

Amongst the 45 patients with urological anomalies, 30 were males and 15 were females. Among the 20 patients with genital anomalies, 15 were males and 5 were females.

Fable 1: Detection of	urinary tract anon	nalies in patients v	with $(n = 75)$
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Investigation	No. of patients	No. of males $(n = 50)$	No. of females $(n = 25)$
Ultrasound examination	35	28	7
VCUG	45	30	15

Urological anomaly	No of Patients	
Vesico-ureteric reflux	28%	
Renal agenesis	5%	
Unilateral ectopic kidney	2%	
Duplication	2%	
Crossed ectopia with fusion	1%	
Posterior urethral valves	6%	
Hydronephrosis	10%	
Urethral stenosis	4%	

Table 2: Distribution of urological anomalies

In present study, vesico-ureteric reflux was the most common urological abnormality (28%). Few patients had more than 1 anomaly.

Gender	Anomaly	No. of patients
Males	Cryptorchidism/undescended testis	10
	Hypospadiasis	3
	Bifid scrotum	2
Females	Vaginal agenesis	2
	Bicornuate uterus	3
Total		20

 Table 3: Genital tract anomalies (n=25)

Undescended testis (10%) was the most common genital anomaly, followed by presence of cloaca (5%).

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No of patients with anorectal anomalies	Urological anomalies seen in	Genital anomalies seen in
Males (n=65)	30%	15%
Females $(n = 35)$	15%	5%
Total	45%	20%

Males are the most predominant ones to have uro-genital anomalies. Out of the 65 % of males, 45% had uro-genital anomalies and out of the 35% of females, 20% had uro-genital anomalies.

Discussion

According to few studies, the prevalence of urogenital anomalies in patients with ano-rectal malformations have been observed to be around 26-52% ^[4-6]. Neonates with high ARM's are more prone to develop urogenital anomalies than ones with low anomalies ^[7, 8].

Present prospective study was conducted including 100 neonates with ano-rectal anomalies.

Renal ultrasound examination was able to detect 35% of the urological anomalies, while VCUG was able to detect 45% of the urological anomalies. This is suggestive of the high sensitivity of VCUG.

The incidence of urological anomalies was 45% and that of genital anomalies was 20%. Males were the most common ones to get affected (30% of urological anomalies and 15% of genital anomalies). This observation was similar to the study done by Metts *et al.*, ^[9].

The most common urological anomaly observed was vesico-ureteric reflux (28%) followed by hydronephrosis (10%). According to previous study, the incidence of VUR ranges from 19-47.2%. The incidence of VUR in present study (28%) is similar to that observed by Mishemirani *et al.*, ^[10]. However, a higher incidence of VUR was observed in study done by Metts *et al.*, ^[9] (32%), Boemers *et al.* (27%) ^[11] and Misra *et al.*, (37.5%)=12 ^[9]. Hydronephrosis was the second most common urinary tract abnormality (10%), which is similar to the study done by Mortazavi *et al.*, ^[4]

In present study, 20% of the patients had genital abnormalities (15% in males and 5% in females). Incidence of genital anomalies in present study is higher than that observed by Metts *et al.*, ^[9], Mortazavi *et al.*, ^[4] (16.4% and 15.5% respectively). Cryptorchidism was the most common genital anomaly observed in males and bicornuate uterus was the most common anomaly observed in females. Unlike in present study where females had low ARM, no females had genital anomalies in a study done by Mishemirani *et al.*, ^[10]. The lower incidence of genital anomalies could be attributable to the fact that females having abnormal internal genital organs might have normal looking external genitalia.

Conclusion

Anorectal malformations are associated with numerous other abnormalities of which this study finds a significant associate between ARM with uro-genital abnormalities. Thus, screening for urogenital anomalies must be done using ultrasonography. In case of suspicion, VCUG must be done even in patients with normal renal ultrasound examination. Early identification might prevent urinary tract infections and its related complications.

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Conflicts of Interest: NO conflicts of interest to be declared.

References

- Smith CA, Avansino J. Anorectal Malformations. [Updated 2023 Aug 8]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2024 Jan. Available from: https://www.ncbi.nlm.nih.gov/books/NBK542275/
- 2. Singh MP, Haddadin A, Zachary RB. Renal tract disease in imperforate anus. J Pediatr Surg. 1968;3:337-341
- 3. Wiener ES, Kiesewetter WB. Urologic abnormalities associated with imperforate anus. J Pediatr Surg. 1973;8:151-157.
- 4. Urogenital anomalies associated with anorectal malformations [Internet]. pjms.com.pk. [cited 2024 Jun 29]. Available from: https://pjms.com.pk/issues/aprjun107/article/article4.html
- Bauer SB, Koff SA, Jayanthi VR. Voiding dysfunction in children: neurogenic and non-neurogenic. In: Walsh PC, Retic AB, Vaughan ED, Wein AJ. Campbells Urology. 8th edition. Philadelphia; Saunders; 2002. p. 2250-4.
- 6. Marlinez-Frias ML, Bermejo E, Rodriguez-pinilla E. Anal atresia, vertebral, genital and urinary tract anomalies: A primary polytopic developmental field defect identified through an epidemiological analysis of associations. Am J Med Genet. 2000;13:95(2):169-73.
- 7. Belman BA, King LR. Urinary tract abnormalities associated with imperforate anus. J Urol. 1972;108(5): 823-4.

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- 8. Sangkhathat, S, Patrapinyokul S, Tadtayathikom K. Associated genitor-urinary tract anomalies in anorectal malformation. A thirteen year review. J Med Assoc. Thai. 2002;85(2):289-96.
- 9. Metts JC, Kotkin L, Kasper S, Shyr YU, Adams MC, Brock JW. Genital malformations and coexistent urinary tract or spinal anomalies in patients with imperforate anus. J Urol. 1997;158:1298-300.
- Mirshemirani A, Ghorobi J, Roozroukh M, Sadeghiyan S, Kouranloo J. Urogenital Tract Abnormalities Associated with Congenital Anorectal Malformations [Internet]. [cited 2024 Jun 29]. Available from: https://tspace.library.utoronto.ca/bitstream/1807/58667/1/pe08028.pdf
- 11. Boemers TML, Jong TPVM, Van Gool JD, Bax KMA. Urologic problems in anorectal malformations part 2: Functional urologic sequelae. J Pediatr Surg. 1996;31(5):634-7.
- Misra D, Mushtaq I, Dpake DP, Kiely EM, Spitz L. Associated urologic anomalies in low imperforate anus are capable of causing significant morbidity. J Urol. 1996;48:281-3. 14.