



E-ISSN: 2616-3470
P-ISSN: 2616-3462
© Surgery Science
www.surgeryscience.com
2019; 3(4): 100-103
Received: 18-08-2019
Accepted: 22-09-2019

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International Journal of Surgery Science

A study on Etiology and incidence of formation of types of Anorectal malformation: A prospective study

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DOI: <https://doi.org/10.33545/surgery.2019.v3.i4b.225>

Abstract

Background: Anorectal malformations are one of the most common congenital defects. This study was undertaken to study the hospital incidence of anorectal malformations (ARM), frequency of various types of defects, their sex distribution and the spectrum of anomalies associated with ARM.

Materials and Methods: Ninety consecutive children attending the paediatric surgery department were included in this study. A detailed history was taken, and examination was performed for the primary as well as the associated defects. Appropriate investigations like invertogram, cologram were done wherever indicated. Management was as per the standard protocol. The data was recorded and analyzed.

Results: Out of the 90 patients, 52(57.77%) male patients and 38(42.22%) female patients. Most of our patients presented within first 24 hours of life. Patients who presented after 72 hours were either female patients with anovestibular malformation or male patients with anocutaneous fistula. In the present study, High anomalies are the most common anorectal malformation in this study. Cardiac and Urological abnormalities are the most common associated anomalies with anorectal malformation. In this study we have performed ultra sound in all and voiding cystourethrogram in 87/90 patients. In 29 patients ultrasound was abnormal. VCUG was abnormal in 25 patients.

Conclusions: Anorectal malformations occurred equally in males and females. Females had intermediate defects more frequently, rectovestibular fistula being the commonest. Males were more likely to have high lesions; anorectal agenesis without fistula was the commonest defect. The most common associated defects seen were vesicoureteric reflux and esophageal atresia. Complications were seen more commonly in males with high lesions. There was a significant association between presence of an associated defect and mortality and morbidity.

Keywords: Anorectal, anovestibular, rectovestibular, cystourethrogram

Introduction

Anorectal malformations present with an incidence of 2.0 to 2.5per 10,000 live births¹. These lesions have variable clinical presentations varying from low to complicated high lesions. The low anorectal malformation lesions may be managed by single stage surgical correction, but the complicated or high lesions need to be managed with multi staged operations. These surgical reconstructive procedures are associated with morbidity and mortality. The associated VACTERL (vertebral, anorectal, cardiac, tracheoesophageal, renal, and limb) anomalies if present, further add to the existing morbidity and mortality, associated with surgical reconstruction.

Of these VACTERL anomalies, genitourinary lesions are a frequent cause of significant morbidity and mortality. These genitourinary anomalies include both structural and functional lesions. The genitourinary anomalies occur frequently in patients with anorectal malformation and retrospective reviews report incidence of 20to50% [2].

Improper management of these genitourinary anomalies leads to unnecessary loss of therapeutic outcome and compromised result. This prospective study was done on ninety patients of anorectal malformations who presented to Institute of Government General Hospital / Guntur Medical College, Guntur from September 2014 to March 2017. This study was done to note the association of different types of urogenital anomalies in anorectal malformations."

To evaluate the incidence of association structural and functional genital and urinary anomalies in anorectal malformations. This prospective study was done with an aim to evaluate the incidence of urogenital anomalies associated with anorectal malformations.

Materials and Methods

The present prospective study was done in a single unit on ninety patients with anorectal malformation, who presented for review to Government General Hospital/ Guntur Medical College, Guntur from September 2014 to March 2017. Of these, 52 were male and 38 were female patients. All patients underwent a detailed clinical examination, evaluation and management which were done in the neonatal period. The associated urogenital anomalies were noted.

The anorectal malformations were classified according to Wingspread classification into low, intermediate, and high anomalies.

The radiology workup for every case include X-ray babygram, invertogram, lateral plain X-ray of lumbosacral spine, echocardiography, ultrasonogram of abdomen (Essota color Doppler A.S. machine with 3.5MHz convex probe and 7.5 MHz high resolution linear probe) and VCUg (voiding cystourethrogram), VCUg was done in 87 patients except for 3 patients with cloacal anomaly who underwent genitogram and panendoscopy. Bladder capacity was calculated by the formula weight in Kg x7 = bladder volume in ml Under aseptic precautions a 6Fr infant feeding tube is passed per urethra. Required volume of 76% of urograffin one in three dilutions in required quantity was instilled in to the bladder and a radiograph was taken during micturition.

Grading of VUR (vesicoureteric reflux)

This was done according to international classification.

- Grade I: Reflux into nondilated ureter
- Grade II: Reflux into renal pelvis and calyces without dilatation
- Grade III: Reflux with mild-to-moderate dilatation and minimal blunting of fornices
- Grade IV: Reflux with moderate ureteral tortuosity and dilation of pelvis and calyces
- Grade V: Reflux with gross dilatation of ureter, pelvis, and calyces, loss of papillary impressions, and ureteral tortuosity

The patients with normal ultrasonogram but with reflux on VCUg were noted. All children with VUR and genitourinary malformations were treated by standard protocol. The structural and functional genitourinary anomalies were in particular analyzed in an elaborate manner. The genital anomalies were diagnosed by physical examination, followed by imaging studies when needed. These patients were followed in outpatient department. The observations and results of the patients were tabulated and analyzed.

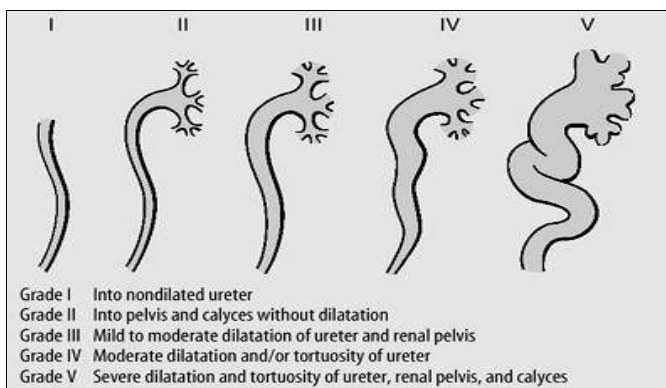


Fig 1: Grading system



Fig 2: Grades of VUR

Observation and Results

From September 2014 to March 2017 90 cases of anorectal malformations were studied and evaluated for associated anomalies. The following parameters were tabulated and analyzed. There were 52 (57.77%) male patients and 38 (42.22%) female patients.

Table 1: Age at presentation

Age (D-Days)	Number	Percentage
<D1	58	64.44%
D2	08	8.89%
D3	08	8.89%
D4	3	3.33%
D5	7	7.78%
D6	0	0%
>D7	6	6.68%

Most of our patients presented within first 24 hours of life. Patients who presented after 72 hours were either female patients with anovestibular malformation or male patients with anocutaneous fistula.

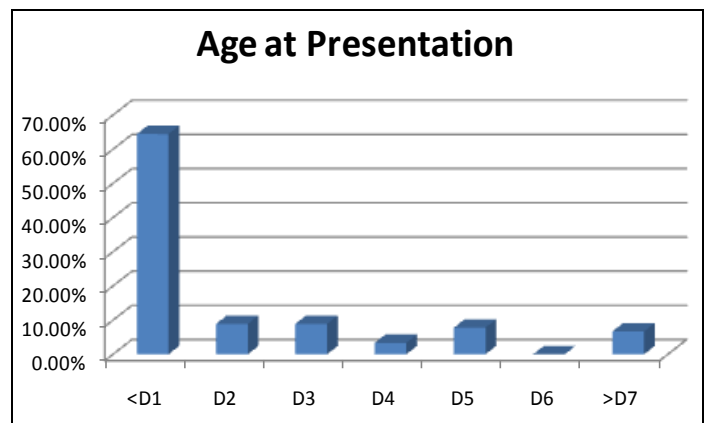


Fig 3: Showing Age of cases

Table 2: Level of anorectal malformation

Level of anorectal malformation	Number	percentage
Low	30	33.33%
Intermediate	22	22.44%
High	38	42.22%

High anomalies are the most common anorectal malformation in this study.

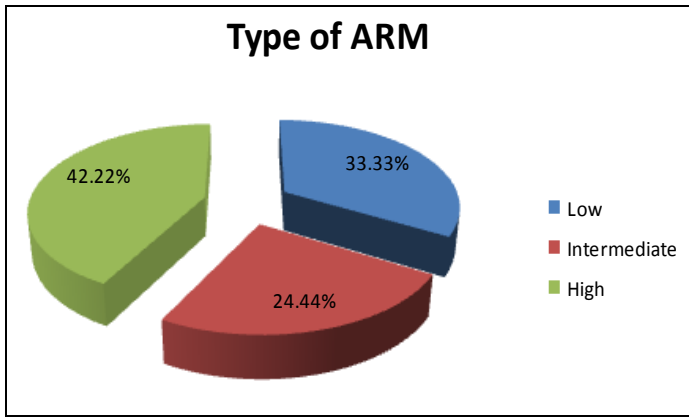


Fig 4: Level of anorectal Malformation

Table 3: Incidence of associated anomalitis

Type of anomaly	Number	Percentage
Urologic	39	43.33%
Cardiac	56	62.2%
Sacrospinal	18	20%
Genital	13	14.44%
Alimentary	08	8.88%
Limbs	05	5.56%

Cardiac and Urological abnormalities are the most common associated anomalies with anorectal malformation. In this study we have performed ultra sound in all and voiding cystourethrogram in 87/90 patients. In 29 patients ultrasound was abnormal. VCUg was abnormal in 25 patients.

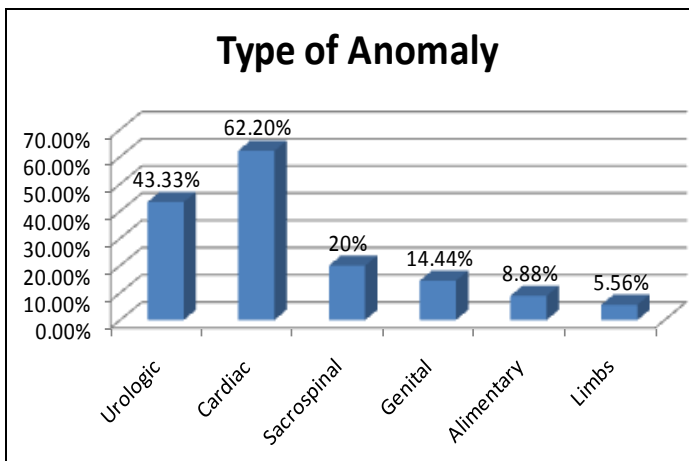


Fig 5: Type of anomaly

Table 4: Urologic anomalies detected in Ultrasound

Type of urologic anomaly (Renal units)	Number
Renal agenesis	4
Renal cysts (Including MCDK)	3
Hydro nephrosis	9
Duplex system	3
Ectopic	3
Hydro uretro nephrosis	20

By Ultrasound 29(42 renal units) had urologic anomalies. 20renal units had hydroureteronephrosis, followed by hydronephrosis in 9 renal units. Other anomalies detected were renal agenesis, dysplastic kidney, ectopia and duplex.

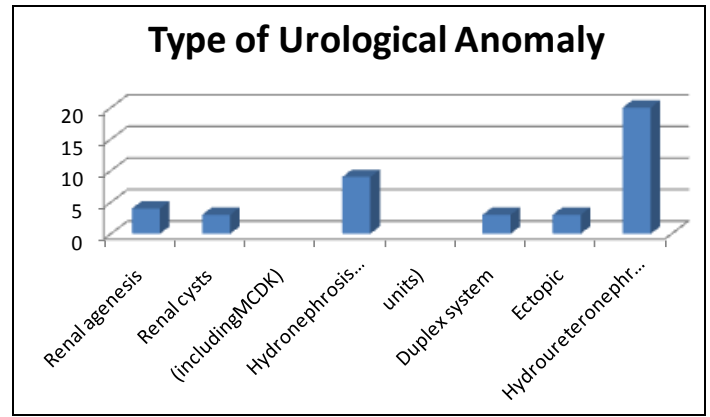


Fig 6: Types of Urologic Anomaly

Table 5: Anomalies detected on VCUG

Type of abnormality	Number
VUR (Renal Units)	38
I	0
II	0
III	1
IV	1
V	36
Posterior urethral Valves	02

38 units of VUR were detected by VCUG. 94.7% were high grade VUR. Grade V VUR was bilateral in 14 patients and unilateral in 8 patients. In 10 patients the ultrasound was normal but MCUG detected VUR. 6 patients with high ARM who had normal ultrasound were detected with VUR. The other 4 patients had Intermediate ARM.

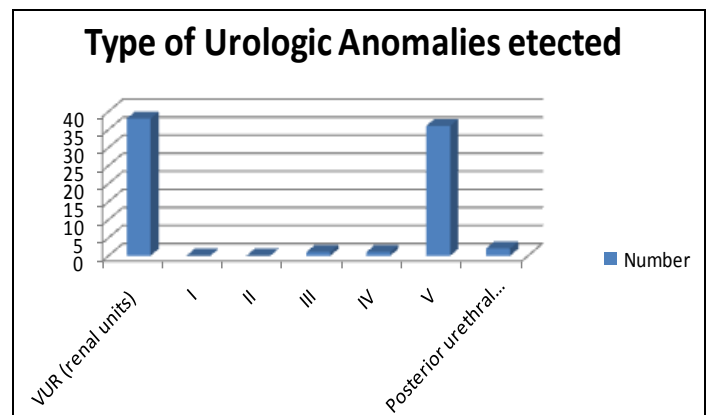


Fig 7: Type of Urologic anomalies detected

Discussion

Urinary tract abnormalities are the most common associated anomaly in patients with ARM and have been reported in 26 to 52% of several large series [1] Its incidence is higher in infants with a high versus a low anomaly and boys are more prone than girls to have an urologic anomalies [2] A Tohda *et al.* found urogenital anomalies in 38.1% of low, 65.5% of intermediate and 85.7% of high ARM. Srivastava *et al* found urogenital anomalies in 16, 2% of low, 22.54% of intermediate and 68.75% of high ARM [3].

About 43.33% of our cases had urinary tract anomalies with preponderance to high level ARM. Our study shows 16.67% of low, 4.44% of intermediate and 22.22% of high ARM to have associated urogenital anomalies.

VUR and renal agenesis are the most common associated urinary tract anomalies with imperforate anus [4, 1]. Associated urologic anomalies in 25.6% and genital anomalies 14% were found. Boemers found VUR in 32% and 27% of their cases respectively [1]. Misra *et al.* reported that 37.5% of patients with low deformity had VUR, but Rattan and Srivastava reported the incidence of VUR only in 1.7% and 5% of their patients [6]. This wide variation in incidence of VUR is related to the differences in the method of evaluation. In some studies VCUG was performed only when sonographic findings were abnormal [5].

In our study USG abdomen and VCUG was performed on all patients. In 43.33% of patients USG was abnormal. VCUG was abnormal in 28.73% of patients; the commonest anomaly detected in our study was VUR. 38 units of VUR were detected by VCUG 94.7% were grade Sin 10 patients the USG was normal but VUR was detected on VCUG. Grade 5 VUR was commonly detected. Six patients with high ARM who had normal USG were found to have VUR [3].

Conclusion

A better understanding of pathogenesis with more accurate pre- and intraoperative diagnosis together with continuous perfectible practice of established operative techniques appear to be the keys to success in ARMs management, aligning our team experience and expertise toward reputed centers. We support a multidisciplinary approach to these cases, using complex teams that include neonatologists, paediatrics, surgeons, paediatricians, psychologist and family doctors, together with a monitored transition towards adult gastroenterology centers.

Acknowledgement

The authors thankful to Department of Paediatrics and Surgery for providing all the facilities to carry out this work.

Conflict of Interest: None

Ethical approval: Taken from Institutional Ethics Committee.

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