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Research Article

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Cardiovascular, Skeletal, Spinal Anomalies Associated with Anorectal Malformations

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ABSTRACT

Introduction: Anorectal malformations are one of the common congenital anomalies with an incidence of 1-3000 to 5000. Up to 70% of the patients have associated anomalies. Urogenital anomalies are the most common associated anomalies followed by cardiac, gastrointestinal and spinal anomalies.

Aim/Objectives: This study was done to determine the type and frequency of non-urogenital anomalies associated with different variants of ARMs according to the Krickenbeck classification.

Materials and Method: 150 patients were included in the study from the Department of Paediatric Surgery, Bangalore Medical College and Research Institute. A full physical examination of the child was conducted followed by infantogram, echocardiogram, spinal ultrasound scan was done to investigate different associated anomalies. MRI was done if further clarity was needed. The patients were classified according to the Krickenbeck classification.

Result: ARM with perineal fistula was the common type of ARM. 73% had associated anomalies, 60.6% had cardiac anomalies including ASD and VSD. 10% had musculoskeletal anomalies and 3.3% had anomalies associated with spine.

Discussion: The presence and severity of associated anomalies is one of the major aspects that determine survival and prognosis. Anomalies like vertebral anomalies may not be life threatening but may have direct impact on the functional outcome of the case, while other anomalies involving gastrointestinal tract and cardio vascular system may lead to mortality and morbidity.

Key Words: Anorectal malformation, Cardiac, Gastrointestinal, Krickenbeck classification, Rectobulbar fistula, Spinal anomalies, Vestibular fistula

INTRODUCTION[1,2]

Anorectal malformation (ARM) consists of a wide range of congenital anomalies which contributes a major share to pediatric surgery admission. ARM affects around 1in 1500 to 1 in 5000 live births.[1] ARM is associated with maldevelopment of neighboring structures arising from caudal cell mass. It includes a spectrum of defects ranging from ones that are easy to treat to those that are more complex and frequently associated with other anomalies.[2] 40-70% of ARM patients have one or more additional defects of other organ systems[1] including urogenital, cardiac, gastrointestinal and spinal anomalies. VACTERL/VATER association is defined by the presence of at least three of the following congenital anomalies: vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies and limb anomalies. In addition to these core components, patients may also have other congenital anomalies. VACTERL association is estimated to occur in approximately 1 in 10000 live births.[3] The presence of associated anomalies is one of the major factors which determine the mortality and clinical outcomes.

MATERIALS AND METHODS

This prospective study includes all patients with ARM who were treated and followed up in the Department of Pediatric Surgery in Bangalore Medical College and Research Institute, Bangalore for a period of three years from Jan 2014 to Dec 2016. This included 150 patients. A detailed clinical proforma was filled for each patient including clinical presentation and physical examination. ARMs were classified according to Krickenbeck classification. Congenital anomalies were also noted.

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A full physical examination of the child was conducted for the evaluation of visible associated anomalies and other abnormalities. A cross table lateral radiograph was performed to identify the level of gas shadow to determine the level of lesion. Infantogram was done to identify skeletal anomalies.

Echocardiogram was done to evaluate the anomalies of the cardiovascular system. Spinal ultrasound scan was done to determine the spinal anomalies. MRI was done if further clarity was needed in the anomaly.

RESULTS

150 patients were included in the study. These patients were classified according to the Krickenbeck classification (Table 1). 101(67.3%) were males and 49(32.6%) were females. Perineal fistula (20%) was the most common type of anomaly present in our study. Vestibular fistula was the frequent anomaly in females while in males it was rectobulbar fistula and no fistula.

Associated Anomalies (Table 2-4)

Associated anomalies were present in 110 (73.3%) patients while 40(26.6%) had isolated ARM. 60.6% of the patients had cardiac anomalies. We included patent foramen ovale (PFO) (40.9%) and patent ductus arteriosus (PDA) (22.7%) in our study even though no active medical or surgical treatment is required for these anomalies. Atrial septal defect (ASD) was found in 35.4% of patients, ventricular septal defect (VSD) in 10.9%. 2 patients had Teratology of Fallot.

10% had musculoskeletal anomalies with sacral agenesis being the commonest. Anomalies associated with the central nervous system was found in 3.3% of patients. VACTERL associated was found in 11 patients.

DISCUSSION

Anorectal malformations are frequently encountered in a wide spectrum of congenital anomalies which pose regular challenges to pediatric surgeons. ARM has an incidence ranging between 1 in 1500 to 1 in 5000.[3,4] Earlier ARMs were classified into high, intermediate and low anomalies. In 2005 a new international diagnostic classification system was devised at the Krickenbeck conference which classified ARM into major clinical group and rare variants.[5] The presence of associated anomalies is one of the key factors which determines morbidity, mortality and long-term functional outcome.

The male female ratio associated with ARM is almost equal.[3,6] But in few variants of ARM male and female preponderance is seen.[5] In the present study, the male female ratio was 2:1. Rectobulbar fistula and ARM with no fistula was the most common type of ARM present in the males, while in females vestibular fistula was common followed by ARM with perineal fistula. Balanenscu et al. reported the presence of 22% of patients with vestibular fistula and 40% with no fistula.[6] Rollins reported the occurrence of perineal fistula in 40.4% males and vestibular fistula in 27.6% which was almost similar to the findings in our study.[7] Stenstrom reported perineal fistula in 46% and vestibular fistula in 18%.[8]

The presence and severity of associated anomalies is one of the major aspects which determine survival and prognosis. Anomalies like vertebral anomalies may not be life threatening but may have direct impact on the functional outcome of the case, while other anomalies involving gastrointestinal tract and cardio vascular system may lead to mortality and morbidity.[9] Almost 50% of ARM may have associated anomalies with an incidence ranging from 30-70%.[6,7,10,11,12] In the present study 73.3% had associated anomalies which is higher than reported literature.

Cardiac malformations have been reported in 30-80% of affected individuals.[13,14] Cardiac malformations may include minor anomalies like vascular anomalies to complex anomalies like Tetralogy of Fallot which may require multiple surgical and long-term medical interventions. Most of the associated cardiac anomalies are treated at a later stage after the repair of the other major anomalies except complex cases with cyanosis. In our series, cardiac anomalies were found in 60.6%. The most common cardiac defects were PFO and ASD. VSD was found in 12 cases while 2 patients had Tetralogy of Fallot. Studies by Rintala and Balanescu show an incidence of cardiac anomalies of 17% and 16% respectively, which is way less than our findings.[3,6] The presence of Tetralogy of Fallot in our study is similar to the findings in previous studies.[6] All variants of ARM in our study had cardiac anomalies in which 75% of patients with rectobulbar fistula had cardiac anomalies.

The reported prevalence of skeletal anomalies found in patients with ARM varies between 20-30%.[3,9,15] In the present series, skeletal anomalies were found only in 9.3% of the patients. Pre-axial limb defects like bilateral thumb agenesis were also found in the present study. Radial agenesis was found in two cases. Quan and Smith in 1973 reported the association of ARM and VATER syndrome, later limb deformities were included to form VACTERL association.[5] VACTERL association has an incidence of 1 in 4000 to 1 in 10000.[7,13] In the author's study 11 cases of VACTERL association is reported. 60-90% patients with VACTERL association will have vertebral anomalies.[16] The vertebral deformities may include defects of one or more than one vertebrae. Common types of vertebral anomalies include hemivertebrae, butterfly vertebrae, vertebral fusions, supernumerary or absent vertebrae. Rib anomalies are also seen along with vertebral anomalies. Abnormal spinal curvatures are also found along with costovertebral defects.

The incidence of tracheoesophageal fistula (TEF) is approximately 1 in 3500.[17] 50-80% of patients with VACTERL association will have TEF. In the present study, only 27.2% of patients with VACTERL association had TEF. Esophageal atresia is also found along with TEF.

Gastrointestinal anomalies are relatively uncommon in ARM patients. The reported literature showed an incidence between 9-24%.[3,6,10,11] In the present study, only 6% patients had gastrointestinal anomalies. The association of Hirschsprung disease (HSD) and ARM is very rare and has been described only in 2.3-3.4% of cases.[18,19,20] In the present study one patient with rectobulbar fistula had HSD.

The association of spinal anomalies with ARM was found to have wide geographical variation. Literatures from the western countries report an incidence up to 46% but in India it is reported to be only 10%.[21] Only 2% of patients in this study had spinal anomalies.

CONCLUSION

Krickenbeck classification of ARM divides ARM into two groups: major clinical groups and rare or regional variants. It is based on the frequency of occurrence and this classification helps in measuring clinical outcomes. Rectobulbar fistula and vestibular fistula was the most recurrently found ARMs. Anomalies related to the cardiovascular system contributes a major share to associated anomalies. Defects in the spine was the least found associated anomaly. These findings push for the need for thorough evaluation of non-urological associated anomalies in ARM patients.

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Authors' Contribution:

Sameer PA: Project conceptualisation, data collection, analysis, writing original draft

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Table 1 Type of ARM: Krickenbeck classification

Type of ARM	Number (%)	Male	Female
Cloaca	7 (4.7)	NA	7
No Fistula	28 (18.7)	28	0
Rectobulbar Fistula	28 (18.7)	28	NA
Rectoprostatic Fistula	21 (14.0)	21	NA
Vestibular Fistula	22 (14.7)	NA	22
Pouch Colon	6 (4.0)	4	2
Rectal Atresia	4 (2.7)	3	1
Rectovaginal Fistula	2 (1.3)	NA	2
Anorectal Agenesis	2 (1.3)	0	2
Perineal Fistula	30 (20.0)	17	13
TOTAL	150	101 (67.3%)	49 (32.7%)

NA – Not applicable

Table 2: Percentage distribution of associated anomalies

Type of ARM	Number	Anomalies Present	
		Number	Percentage (%)
Cloaca	7	7	100.0
No Fistula	28	19	67.9
Rectobulbar Fistula	28	24	85.7
Rectoprostatic Fistula	21	17	81.0
Vestibular Fistula	22	11	50.0
Pouch Colon	6	5	83.3
Rectal Atresia	4	3	75.0
Rectovaginal Fistula	2	2	100.0
Anorectal Agenesis	2	1	50.0
Perineal Fistula	30	18	60.0

Table 3: Frequency of associated anomalies with ARM

Type of ARM	Number	CVS (%)	Musculoskeletal* (%)	CNS (%)	GIT (%)
Cloaca	7	5 (71.4)	4 (57.1)	1 (14.3)	1 (14.3)
No Fistula	28	17 (60.7)	2 (7.1)	2 (7.1)	1 (3.6)
Rectobulbar Fistula	28	21 (75.0)	3 (10.7)	1 (3.6)	0 (0.0)
Rectoprostatic Fistula	21	15 (71.4)	2 (9.5)	1 (4.8)	2 (9.5)
Vestibular Fistula	22	11 (50.0)	0 (0.0)	0 (0.0)	3 (13.6)
Pouch Colon	6	3 (50.0)	2 (33.3)	0 (0.0)	0 (0.0)
Rectal Atresia	4	1 (25.0)	1 (25.0)	0 (0.0)	1 (25.0)
Rectovaginal Fistula	2	2 (100.0)	0 (0.0)	0 (0.0)	0 (0.0)
Anorectal Agenesis	2	1 (50.0)	0 (0.0)	0 (0.0)	0 (0.0)
Perineal Fistula	30	15 (50.0)	1 (3.3)	0 (0.0)	1 (3.3)

CNS = central nervous system; CVS = cardiovascular system.
*Includes craniofacial anomalies

Table 4: Anomalies associated with ARM

System	Anomaly	Number
Cardiovascular	PFO	45
	ASD	39
	PDA	25
	PAH	16
	VSD	12
	Dilated Right side Chamber	10
	TR	8
	Tetralogy of Fallot	2
	Coarctation of Aorta	2
	Aortic Sinus Rapture	1
	Dilated Coronary Sinus	1
	Interrupted Aortic Arch	1
	Levocardia	1
	Double Outlet of Right Ventricle	1
	Mitral Valve Prolapse	1
Skeletal (Including Craniofacial)	Sacral Agenesis	3
	Radial Agenesis	2
	Butterfly Vertebrae T3	2
	13th Rib	1
	Right Thumb Agenesis	1
	Bifid L5, S1	1
	Bilateral Thumb Agenesis	1
	Bilateral CTEV	1
	Cleft Palate	1
	Fused L3 , L4	1
	Fused L5 , L6	1
	Fused S1 , S2	1
	Hemi Sacrum	1
	Hemi Vertebrae L3	1
	Hemi Vertebrae T4, T5	1
	Hemi Vertebrae L4	1
	Pelvic Diastasis	1
	Retrognathia	1
	Scoliosis	1
	Supernumerary L6	1
Spinal	Low Lying Spinal Cord	2
	Partial Segmentation	1
	Lesion in Conus Medullaris	1
Gastro Intestinal	TEF	4
	Esophageal Atresia	2
	HSD	1
	Biliary Atresia	1
	Duplicated Distal Pouch	1
Others	Omphalocele	2
	Inguinal Hernia	1
	Lumbar Hernia	1
	Omental Cyst	1
	Noncommunicating Hydrocephalus	1
	Facial Nerve Palsy	1