Original Article

Anorectal Malformations and Their Impact on Survival

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Abstract. ***Objective :*** To evaluate the incidence, types and the effect on outcome of associated anomalies in neonates with anorectal malformations (ARM). ***Methods :*** This retrospective study was carried out on all neonates with ARM admitted to the neonatal surgical intensive care unit (NSICU) from 1998 through 2003. ***Results :*** Of the 754 neonates admitted to the NSICU during the study period of 6 years, there were 124 (16.4%) neonates with anorectal malformations. Of these 110 were included in the study. 73 % were male and 27% female. 86% of these were high ARM (HARM) while only 14% were low ARM (LARM). Associated anomalies were seen in 68% of patients. The incidence was 72% for HARM and 50% for LARM. The major associated anomalies consisted of esophageal (13%), gastrointestinal (GIT) (11%), genitourinary (GUT) (32%), skeletal (26%), cardiac (33%) and miscellaneous 26%. The overall survival rate was 84% (82% for HARM and 94% for LARM). The survival among those with associated esophageal anomalies was 43%, GIT 67%, GUT 80%, cardiac 61%, skeletal 76% and miscellaneous 79% respectively. This difference in survival was significant only for those with esophageal (p=0.004) and cardiac anomalies (p=0.0026). The survival rates among those with one, two or more than two organ systems involved with associated anomalies were 88%, 82% and 58% respectively. This difference was significant only for more than two organ systems involvement (p=0.003). ***Conclusion :*** Associated anomalies are common in neonates with ARM, the incidence being similar for HARM and LARM. The survival depends upon the number and severity of associated anomalies both in patients with LARM and HARM. Neonates with more number of organ systems involved have a poorer survival specially when associated with esophageal and cardiac anomalies. All neonates with ARM merit a meticulous search for associated anomalies so that the management can be tailored for each baby. **[Indian J Pediatr 2005; 72 (12) : 1039-1042]** *E-mail : sandpagr@hotmail.com*

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Malformations of other organ systems in patients with anorectal anomalies have been variously reported. Their association has profound impact on survival. The incidence of associated organ system anomalies depends upon the meticulousness with which they have been searched for by clinical examination and investigative procedures. The incidence is variously reported from 30 – 70%,[1-8] some being minor anomalies but others being life threatening. Though abnormalities of all system have been described, genitourinary (GUT), cardiovascular, gastrointestinal (GIT) and vertebral anomalies are the ones commonly reported. There is paucity of literature describing the effect of associated anomalies on the survival of neonates with ARM during the initial management. This retrospective study describes the incidence of associated anomalies and its effect on survival in neonates with ARM admitted to the neonatal surgical intensive care unit (NSICU) of a tertiary care center in a developing country.

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Abbreviations

ARM	=	Anorectal malformation
HARM	=	High anorectal malformation
LARM	=	Low anorectal malformation
NSICU	=	Neonatal surgical intensive care unit
GIT	=	Gastrointestinal tract
GUT	=	Genitourinary tract
EA	=	Esophageal Atresia
TEF	=	Tracheo-esophageal fistula

MATERIALS AND METHODS

This retrospective study includes all the neonates with ARM admitted in the NSICU, All India Institute of Medical Sciences from January 1998 through December 2003. The investigations done at the time of admission were a babygram (X-ray of the whole baby including chest, abdomen and limbs), an invertogram, ultrasound abdomen and an echocardiogram. A routine MCU was not done in the initial work-up of these neonates but was planned to be done later in follow-up with the distal cologram, prior to definitive surgery. Case records of all these cases were retrieved and evaluated for types of ARM, the associated anomalies and the outcome. For practical management strategies, the intermediate

anomalies have been clubbed with the high ARM and analyzed as such. Data was analyzed and their impact on survival was studied using program SPSS 11.3 for Windows. Univariate as well as multivariate logistic regression analysis was performed and the risk factors of survival were calculated. P value equal to 0.05 or less was considered as statistically significant.

RESULTS

In this study period of 6 years, 754 neonates were admitted to the NSICU of which 124 (16.4%) cases were diagnosed to have ARM. Of these 124, only 110 cases have been included in the present study, as the record of 14 cases could not be retrieved. Thus, 110 neonates with anorectal malformation (ARM) have been the subject of the present study. There were 80 (72.7%) males and 30 (27.3%) females.

Of these 110 neonates, 85.5% (94 of 110) had high anorectal malformation (HARM) and 14.5% (16 of 110) had low anorectal malformation (LARM) (Table 1). Amongst these 94 neonates with HARM there were 23.4% (22 of 94) cases of pouch colon (congenital short colon). These 22 pouch colon cases comprised of type I (22.7%), (9.1%) type II, and (68.2%) type IV. There were no type III pouch colon. Five of these 22 babies with pouch colon also had common cloaca. In addition, there were eight more cases of common cloaca making the total number of babies with common cloaca to be 11.8% (13 of 110) of ARM. Associated anomalies were detected in 68.1% (75 of these 110) neonates with ARM. The distribution of HARM and LARM and the incidence of associated anomalies and the survival rates in these are depicted in table 1.

The major associated anomalies consisted of esophageal atresia (EA) with or without tracheoesophageal fistula (TEF) in 12.7% (14 cases), genitourinary anomalies in 31.8% (35 cases), skeletal 26.4% (29 cases), cardiac 32.7% (36 cases), gastrointestinal 10.9% (12 cases) and miscellaneous 26.4% (29 cases) (Table 2). List of associated anomalies detected is depicted in table 2.

Ninety-two babies (83.6%) out of 110 survived which included sixty seven (83.7%) of 80 males and of the 30 females 25 (83.3%). The incidence of number of organ systems involved with associated anomalies and the survival rates in these groups is depicted in Fig. 1.

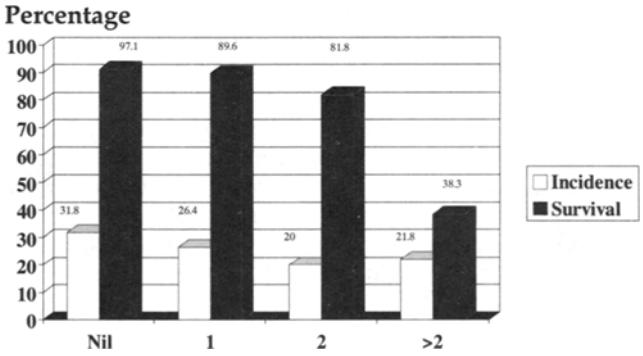

Fig. 1. Incidence of associated anomalies and survival according to the number of organ systems involved

Table 1. The Incidence of Associated Anomalies and Their Survival Rates amongst Males and Females with HARM and LARM.

Type of ARM	Males (n=80)		Females (n=30)		Total (n= 110)	
	Incidence (%)	Survival (%)	Incidence (%)	Survival (%)	Incidence (%)	Survival (%)
HARM	72 (90)	59 (81.9)	22 (73.3)	18 (81.8)	94 (85.5)	77 (81.9)
Anomalies present	52 (72.2)	40 (76.9)	15 (68.2)	11 (73.3)	67 (71.3)	51 (76.1)
Anomalies absent	20 (27.8)	19 (95)	7 (31.8)	7 (100)	27 (28.7)	26 (96.3)
LARM	8 (10)	8 (100)	8 (26.7)	7 (87.5)	16 (14.5)	15 (93.7)
Anomalies present	3 (37.5)	3 (100)	5 (62.5)	4 (80)	8 (50)	7 (87.5)
Anomalies absent	5 (62.5)	5 (100)	3 (37.5)	3 (100)	8 (50)	8 (100)

Table 2. The Incidence of Various Organ Systems Involved with Associated Anomalies and Survival in Cases of HARM and LARM

Organ system Involved with associated anomalies	HARM (n= 94)		LARM (n=16)		Total (n= 110)	
	Incidence (%)	Survival (%)	Incidence (%)	Survival (%)	Incidence (%)	Survival (%)
Esophageal	11 (11.7)	4 (36.4)	3 (18.8)	2 (66.7)	14 (12.7)	6 (42.9)
Gastrointestinal	12 (12.8)	8 (66.7)	0 (0)	0 (0)	12 (10.9)	8 (66.7)
Genitourinary	32 (34)	25 (32)	3 (18.8)	3 (100)	35 (31.8)	28 (80)
Skeletal	24 (25.5)	18 (75)	5 (31.3)	4 (80)	29 (26.4)	22 (75.9)
Cardiac	33 (35.1)	20 (60.6)	3 (18.8)	2 (60)	36 (32.7)	22 (61.1)
Miscellaneous	27 (28.7)	21 (77.8)	2 (12.5)	2 (100)	29 (26.4)	23 (79.3)

DISCUSSION

Associated congenital anomalies in neonates with anorectal malformation assume significance, as survival and prognosis depends upon the number and severity of the associated anomalies. Some anomalies like those of the vertebra, though not lethal, may have a direct bearing on the ultimate functional outcome of the case. Other anomalies involving the cardiac, GIT and GUT may lead to morbidity and mortality during the initial management of neonates with ARM. Various studies have reported the incidence of associated anomalies with ARM to be 30 to 70% of cases.[1-8] (Table 3). The incidence of associated anomalies basically depends upon the meticulousness with which they have been sought for. More often than not, these neonates undergo the initial surgical treatment without detailed work-up for associated anomalies soon after their admission. In the present study 68% of the neonates had one or more organ systems involved with associated anomalies, with nearly one-fifth having more than two organ systems involved in addition to ARM.

The incidence of associated anomalies in relation to HARM and LARM as reported by others is depicted in table 3[9-13], which highlights the higher incidence of associated anomalies in LARM in the present study. The incidence of associated anomalies in HARM and LARM was not statistically different (p = 0.62) in the present study.

Urogenital anomalies in association with ARM have been reported to occur in 38–74%.[1,3,6,12,14-17] (Table 4). This variation depends upon the different methods employed to study the urogenital tract. Genitourinary anomalies have been reported to be more common in the patients with high anorectal malformation. The urinary system anomalies comprise of hydronephrosis, agenesis of kidney, structural and positional variations of the kidneys, pelviureteric junction obstruction and vesicoureteric reflux, whereas cryptorchidism, hypospadias, ambiguous genitalia, bifid scrotum are the common genital anomalies associated with anorectal malformation.[18,19] In the present study, overall incidence of genitourinary tract (GUT) anomalies was 31.8% (35 of 110), with 34% incidence in HARM (32 of 94 cases) and 18.7% in LARM (3 of 16 cases) (Table 2). Some workers have recommended a routine MCU in all patients with ARM, however this has not been the practice in our unit. The MCU is not done in these neonates and is planned at a later date. This may be the reason for lower incidence of detection of VUR in the present study.

The incidence of cardiovascular anomalies reported varies from 17–27%.[3,6,7,12] The most common cardiac anomalies reported are tetralogy of Fallot, followed by ventricular septal defect. These are the anomalies that have most profound impact on the immediate survival of the patient. The incidence of cardiac anomalies in the present study *i.e.*, 32.7% (36 of 110) (Table 2) is higher than what is reported and is probably because of routine echocardiography being done in all cases admitted to this neonatal surgical ICU. The incidence was found to be similar for both HARM and LARM cases (60.6% *vs* 60%) (Table 2), which is against the standard belief that the incidence of cardiac anomalies is less in LARM.

Vertebral anomalies associated with ARM have been reported to be between 26 – 57%.[3,16,20,21] In the present study the incidence of skeletal anomalies constituted 26.4% (29 of 110 cases) (Table 1). This incidence was found to be similar in HARM and LARM (75% *vs* 80%) (Table 2).

A spectrum of gastrointestinal anomalies has been described in cases of anorectal malformation. The reported incidence is between 9 and 24% (Table 4). The most common of the associated gastrointestinal anomaly is esophageal atresia with tracheoesophageal fistula with a reported range of 5 – 10%;[1,22,23] duodenal atresia and/or malrotation varies from 1 – 2%,[3,7] Hirschsprung's disease 2.3% – 3.4%,[22,23] focal ectasia of the terminal bowel with segmental dilatation of colon,[24] pyloric atresia.[25] In the present study GIT (including esophageal atresia) anomalies were noted in 24% (26 of 110 cases).

In the present study the overall survival of neonates with ARM was 83.6%. Though the survival of neonates with HARM was lower than those with LARM (81.9% *vs* 93.8%), this difference was not statistically significant (p = 0.41) (Table 1).

Survival among babies with associated esophageal anomaly was only 42.9% (6 of 14 cases). This difference in survival as compared to those without the anomaly was found to be significant (p = 0.004). The difference in the survival of those with associated cardiac anomalies

Table 3. Incidence of Associated Anomalies According to the Types of ARM as Reported in Literature

Author	Year of publication	Incidence of associated anomalies(%)	
		LARM	HARM
Partridge *et al*[9]	1962	25	62
Swenson *et al*[10]	1967	24	52
Saeki *et al*[4]	1985	31	56
Smith *et al*[5]	1986	40	85
Boocock *et al*[11]	1987	36	80
Mittal *et al*[12]	2004	37	78
Chalapathiel *et al*[13]	2004	5.4	41
Present Study	2004	50	72

Table 4. Reported Incidences of Organ System Involved with Associated Anomalies with ARM

Series	Year	n	GUT(%)	Skeletal(%)	GIT(%)	CVS(%)
Saeki M[4]	1985	345	21	17	9	6
Smith D[2]	1988	246	27	26	10	9
Hassink EA[3]	1996	264	43	38	24	21
Nazer J[6]	2000	54	43	26	–	19
Cho S[7]	2001	103	49	43	18	27
Kos M[8]	2001	125	63	26	17	65
Present Study	2004	110	32	26	24	33

(61.1%; 26 of 36) as compared to those without cardiac anomalies was also found to be statistically significant (p = 0.026). The difference in survival among babies with associated GIT anomalies (66.7%), GUT anomalies (80%) and skeletal anomalies (75.9%) was not found to be significant as compared to those without these anomalies.

The incidence of pouch colon (congenital short colon) in the present study was 20% (22 of 110), and among these, 63.6% (14 of 22) had associated anomalies. Of these patients with associated anomalies, 85.7% (12 of 14) survived. The incidence of common cloaca in the present study was 11.8% (13 of 110), and among these 76.9% (10 of 13) had associated anomalies. The survival rate among patients of common cloaca with associated anomalies was 90% (9 of 10).

The survival rate among neonates with no associated anomaly, and in one or two organ systems involvement was 97.1%, 89.6%, and 81.8% respectively. This difference was statistically not significant. The survival among neonates with more than two organ systems involved with associated anomalies was only 58.3%, which was statistically significantly lower (p=0.003).

The present study reveals that there is high incidence of associated anomalies with ARM and also that there is no significant difference in the incidence of associated anomalies in neonates with HARM and LARM. Further it also shows that with increasing number of organ systems involved, the survival rate decreases but it is significantly low only when more than two organ systems are simultaneously involved with associated anomalies in addition to the ARM. The association of esophageal and cardiac anomalies also independently affects the survival of these babies with ARM. The high incidence of associated anomalies and their effect of survival reinforce the view that these babies need to be adequately investigated during the neonatal admission and their management individualized.

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