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Research Paper

Anorectal Malformations-An Experience from a Tertiary Referral Hospital

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Abstract

Objectives: *This study was undertaken to find out various types anorectal malformations in local population, their sex distribution and compare that with available present day data, to study the incidence of various associated anomalies and to study the morbidity and mortality associated with it.*

Study Design: *70 consecutive patients of anorectal malformation attending the Department of Surgery, Gajra Raja Medical College, Gwalior, India were included in the study. A detailed history was taken and thorough examination was performed for both the primary and associated anomalies. Necessary investigations were done. Management was done according to the protocol. The data was recorded and analyzed.*

Results: *Out of 70 patients 51 were males and 19 were females. Types of malformations were anorectal agenesis without fistula (17.14%), perineal membrane/fistula (15.71%), recto-vestibular fistula (11.43%), recto-urethral and recto-vescical fistulae (7.14% each) and rare anomalies like cloacal malformations (5.71%) and congenital pouch colon(8.57%). 49.01% of males and 15.79% females had associated anomalies. Urogenital system was involved in 15.72%, cardiovascular and skeletal systems in 10% each, respiratory system and gastrointestinal system in 7.14% each and nervous system in 4.29% of patients. There were 11 deaths and 6 of those had severe associated anomalies.*

Conclusions: *Anorectal malformations occurred commonly in males. High anomalies in males and intermediate anomalies in females were common. Commonest anomalies were rectovestibular fistula in females and anorectal agenesis without fistula in males. Urogenital anomalies were most commonly associated. Mortality was higher in males having high anomalies and in patients having severe associated anomalies.*

Introduction

Anorectal malformations (ARM) are relatively frequently encountered anomalies that represent an important component of pediatric surgical practice. They include spectrum of diseases from simple anal membrane to complex anomalies. ARM has an incidence rate ranging from 1:1,500 to 1:5,000 live births, slightly more common in males.[1] In India exact incidence is not available since not many studies are available on the topic.[2]

There is a wide spectrum of possible associations of ARM with various systemic malformations. Urogenital association is most commonly seen, followed by vertebral or spinal.[3] Varieties of syndromes are also associated with ARM namely Down's syndrome, VACTERL or VATER association, Cat-eye syndrome etc.

Previously most commonly used Wingspread classification divided ARM into high, intermediate, low and miscellaneous anomalies. In 2005 new international classification was introduced during the meeting in Krickenbeck Castle in Westphalia, Germany. It divides ARM into two main groups, "major clinical groups" and "rare/regional variants" and is based on frequency of occurrence and allows management outcomes to be measured.[4]

In India numerous studies have showed high prevalence of the disease. Concept of 'pouch colon' is also important with regard to the subcontinent. It is more common in Northern India including Gwalior region where the current study was done.[5]

This study was undertaken to find out various types anorectal malformations in local population, their sex distribution and compare that with available present day data, to study the incidence of various associated anomalies and to study the morbidity and mortality associated with it.

Materials and Methods

This study was conducted in the department of surgery, Gajra Raja Medical College, Gwalior [Madhya Pradesh state, India] during the period September 2013-august 2014. All the neonates those were admitted to the newborn unit diagnosed with anorectal malformation were included in the study. Those who admitted as follow up were also included forming a study group of 70 patients.

A detailed case history was recorded on admission. Special attention was given to the pregnancy history to detect any complications during pregnancy like medications during first trimester, radiation and infectious diseases and to whether the baby was term or preterm. Follow up patients were enquired about the type of procedure done and if there were any complications. Congenital malformation in the family and relatives was also looked for. Physical examination, which included a detailed head to toe examination to look for any congenital defects and a detailed local examination, was done. Perineum was examined for anal membrane or fistula. In female baby vestibule and vagina were inspected for rectovestibular and rectovaginal fistulas. Urine was examined for meconium stain. Neonates were examined thoroughly for associated anomalies with special focus on genitalia, spine and CVS.

Blood and urine examination was done routinely for all cases and neonates with a suspected high or intermediate anomaly were advised erect X-ray abdomen. A USG abdomen was done in all to rule out renal anomalies. Those having cardiac murmur or skeletal deformity were investigated with 2-D ECHO or skeletal X rays respectively. Distal loop cologram (DLC) was done in patients admitted for second stage procedure following initial colostomy. Patients were managed according to the type of anomaly. All the patients were followed up during the period of study in the out-patient department. Special care was taken to assess the neo-anus patency and functioning.

Observations

Out of the 70 babies 7 were born preterm and 63 were term. Low birth weight (LBW) babies constituted 40 out of which 14 were very low birth weight (VLBW). Out of the 70 cases 51 patients were males and 19 were females. Out of the 19 female patients 11 had intermediate anomalies and 2 each had high and low anomalies. The rest were rare anomalies which included 4 cloacal anomalies. Out of 51 male patients 9 were low anomalies, 5 intermediate anomalies and 30 had high anomalies. 7 had rare anomalies. Patients were classified into the Krickenberg groups as given in table1.

All the patients were examined and investigated for associated anomalies. 49.01% of males and 15.79% females had associated anomalies. Associated anomalies were common among males and those with high/intermediate anomalies. Table 2 shows the frequency of anomalies with regard to different systems and individual anomalies involved.

Management was done according to the type of anomaly. All the patients with low anomalies except 3 underwent anoplasty regardless of sex. 2 male patients died in the immediate newborn period due to complications associated with trecheo-esophageal fistula. One patient had associated ileal atresia and had iloestomy. Out of the 8 patients with recovestibular fistula 5 underwent cut back anoplasty and 3 had single stage primary Anterior Sagittal Anorectoplasty (ASARP). Sigmoid colostomy was done for 2 out of 3 patients having rectovaginal fistula and the other underwent primary ASARP. All the patients with high anomalies had either a sigmoid or transverse colostomy done except for 2 for whom ileostomy was done because of associated ileal atresia.

Out of the 4 cases of cloacal anomalies, 3 presented in neonatal period and underwent sigmoid colostomy. One had undergone colostomy in past and during the study period was admitted for second procedure in the form of Posterior Sagittal Anorectoplasty (PSARP). There were 6 cases of congenital pouch colon, all of them males. 4 of them underwent colostomy using the same loop of colon. Remaining 2 had ileostomy done and one of which underwent ASARP during the period of study and also had a rectovescical fistula.

Outcome following a definitive procedure was monitored, both in low and high/ intermediate anomalies. In 8 patients with low anomaly who had undergone anoplasty, 7 had good outcome during the follow up in the study period. A patient presented with anal stenosis later. The complications after colostomy like excoriation, bleeding, constipation or prolapsed were not included in the study. Out of 10 patients with high/intermediate anomalies who had a definitive procedure during the study period, males and females were 5 each. All of them had good outcome except for a female who had cloacal anomaly and 2 males of which one presented with anal stenosis and the other with intestinal obstruction.

11 patients in our series died (10 males and 1 female). The female patient had rectovestibular fistula and had undergone ASARP and was readmitted later with diarrheal disease and succumbed to it. All the males had high anomalies except two, both of them having associated TOF and lost their lives due to it's complications. Of the remaining 8 male babies who died were 2 preterm babies with low birth weight. 1 had associated TOF, 1 had CHD, 1 had associated nervous system anomalies and 1 had Down's syndrome and associated complications, all of them died in early neonatal period. One male baby underwent colostomy, but died on first postoperative day because of medical reasons. There was another baby who was admitted for third stage procedure and had undergone a colostomy closure, but baby died postoperatively due to aspiration during a convulsive episode.

Table 1- classification of patients according to krickenbeck clinical groups

Clinical groups	No of patients	Percentage of total
anorectal agenesis without fistula	12	17.14%
Perineal fistula/ membrane	11	15.71%
Rectovestibular fistula	8	11.43%
Congenital pouch colon	6	8.57%
Rectovescical fistula	5	7.14%
Rectourethral fistula	5	7.14%
Cloacal anomalies	4	5.71%
Rectovaginal fistula	3	4.23%
Anal stenosis	0	0

Table 2- different systems and individual congenital anomalies associated with ARM

System involved	Frequency of anomaly	Individual anomaly	No of cases
Genito-urinary	**15.72%**	Hypospadiasis	**5**
		Hydronephrosis	**2**
		epispadiasis	**1**
		Absent testis	**2**
		Bifid scrotum	**1**
		Exstrophy bladder	**1**
Nervous system	**4.29%**	Meningomyelocele	**2**
		Hydrocephalus	**1**
CVS	**10%**	ASD	**2**
		VSD	**2**
		CHD	**2**
Gastro-intestinal	**7.14%**	Ileal atresia	**3**
		double appendix	**1**
		Omphalocele	**1**
Skeletal	**10%**	Sacral anomaly	**3**
		CTEV	**2**
		Absent radius	**1**
		Flat foot	**1**
Respiratory	**7.14%**	TOF	**5**
Down's syndrome			**1**

Table 3- showing Mortality in Each Sex and type of Anomaly

Type of Anomaly	Male	Female	Percentage
High	8	0	25%
Intermediate	0	1	6.25%
Low	2	0	8.18%

Table 4 incidence of different type of Anomaly in both sexes compared to previous studies

	Sex	High	Inter-mediate	Low	Rare
Stephen and smith[9] (n=260)	Male	52%	0%	48%	0%
	Female	65%	0%	35%	0%
Childrens Memorial hospital[8] (n=216)	Male	50%	10%	40%	0%
	Female	5%	15%	59%	21%
Japanese study group[10] (n=1992)	Male	36%	13%	48%	0
	Female	13%	7%	71%	0%
Our series (n=70)	Male	58.83%	9.80%	17.65%	13.72%
	Female	10.53%	57.89%	10.53%	21.05%

Discussion

Indian incidence of ARM is not available since not many studies are available on the topic in this part of the world.[2] Our series included sporadic referrals from around the region, from different districts and also children presenting later in life. Therefore incidence estimation was not possible.

Most series have reported a higher incidence of ARM among males. Stephens and Smith (1971) reported a male preponderance of 65% in his cases, but collected data from 35 sources showed a male incidence of 57%.[6] The Alberta congenital anomalies surveillance system 1990-2004 shows an overall rate with a marked male predominance (1.7:1).[7] Our study has 51 males to 19 females which equates to (2.68:1).

Low anomalies are by far the commonest of all anomalies noted by most authors. In the children's memorial hospital series of 216 patients 47% had low anomalies, in Liverpool series 74 out of 151 patients had low anomalies.[8] In contrast our series has a high incidence of high anomalies especially in males, with low anomalies constituting only 11 out of 70 patients.

A high incidence of low anomalies has been noted by most authors in both sexes, but low lesions are common in females and high lesions are commoner in males. In our series there were 58.83% high anomalies, 9.8% intermediate anomalies, 17.65% low anomalies and 13.72% rare anomalies in males. Females had 10.53% high, 57.89% intermediate, 10.53% low and 21.05% rare anomalies (table 4). Possible reason for low incidence of low anomaly in our series may be the fact that our centre being a tertiary referral hospital, low anomalies like anal stenosis, anteriorly placed anus etc are managed in district

hospitals and other peripheral centers and are seldom referred to us.

Cloacal anomalies are described in the literature as a very uncommon type of ARM with incidence of 1:250,000 live births.[11] But this study had a greater frequency of cloacal malformations with 4 cases constituting 5.71% of the total.

Congenital pouch colon, which is associated with ARM, is much more common in North India than in the rest of the world. Indian patients account for 92% of all reported cases and pouch colon syndrome accounts for 6–13% of all ARM in Northern India.[5] Our series had 6 cases of the condition constituting 8.57% of all cases.

Associated anomalies are more common in boys (52–63%) and the higher the ARM, the higher the risk of associated anomalies.[12] Our study also had a high incidence in males (49.01%) and high anomalies. Genitourinary anomalies occur in 21–61% of patients. Up to 26% of boys have genitourinary problems; as opposed to 5% of girls.[13] Incidence in our study was 15.72% for genito-urinary anomalies. Vertebral and spinal anomalies are commonly associated, especially with supralevator lesions, with an incidence of 4.6–40%.[10] Our study had an incidence of 14.29% for skeletal and spinal anomalies. Gastrointestinal anomalies are less common, with reported incidence of 10–25%, the commonest deformity being tracheoesophageal fistula (13%) followed by duodenal atresia.[13] There was 7.14% gastro intestinal anomalies other than TOF ,which alone had an incidence of 7.14%. Cardiac malformations occur in 9–20% of defects equally in patients with high and low lesions, with tetralogy of Fallot being the commonest diagnosis.[5] CVS anomalies were more frequent in our study (10%). The general principle is that higher the type of ARM, more severe the associated anomaly which goes the same with our series.

Most authors have recorded a high mortality in patients with ARM in earlier days in the west. Whereas the mortality in the west has reduced substantially due to better neonatal care, but in India it continues to haunt and frustrate people dealing with this condition. In our series 11 out of 70 patients have died. Among them were 2 preterm babies with low birth weight. 6 patients died due to complications associated with associated anomalies. High mortality is associated with high anomalies. Patridge and Gough have reported 35.9% mortality in high anomalies compared to 5.7% in low anomalies. Stephens has reported 45.6% mortality in patients with high anomaly and 22% in those with low anomaly.[9] Our series reports 25% mortality among patients with high, 6.25% in intermediate anomalies and 18.18% in low anomalies. Mortality is common among male patients (19.6%) when compared to females (5.26%).

Conclusion

ARM continues to be a common problem as population grows in the developing world, especially in Indian subcontinent. A lot of research and ways of correcting them have been proposed, but still about 30-40% of these cases require long term services in terms of physiotherapy, daily enemas or sometimes even permanent diapers.

It is well understood that good results can be achieved by properly managing these babies in neonatal period with minimal insult to the sphincters. Present study therefore is an initial experience from a teaching hospital in developing world, results of which are compared to any international standard. Still further long term studies are needed to assess the morbidities like level of continence in patients of ARM.

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