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Anorectal malformations in neonates

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ABSTRACT

Background: Anorectal malformations (ARM) are associated with congenital anomalies and other risk factors, yielding a poor prognosis, especially in neonatal life. **Objectives:** This study was performed to identify the congenital anomalies as a factor of poor prognosis (mortality) in such patients. **Settings:** Department of Pediatric surgery, The Children's Hospital and The Institute of Child Health, Lahore. **Design:** Prospective observational study, with statistical support. **Materials and Methods:** The information on the demography, clinical features, investigations, management performed, and outcome was entered in the designed proforma and analysed with the help of statistical software *EpiInfo* version 3.5.1. Statistical test: Chi-square test was used to determine statistical significance of the results. **Results**: Of 100 neonates with ARM, 77 were male and 23, female (3.4:1). The mean age at presentation was 3.4 days (range, 12 hrs to 28 days). In 60 patients (60%), the presentation was imperforate anus without a clinically identified fistula. In 28 patients (28%), associated anomalies were present. The common associated anomalies were urogenital (10%), cardiovascular (8%), and gastrointestinal (6%). Down's syndrome was present in 8 (8%) patients. A total of 15 (15%) deaths occurred in this study. In patients having associated congenital anomalies, 11 deaths occurred, whereas, 4 deaths were in patients without associated anomalies ($P < 0.5$). **Conclusion**: The mortality is higher in neonates with ARM having associated congenital anomalies.

Key words: Congenital anomalies, imperforate anus, mortality, neonate

INTRODUCTION

In the past, the mortality rate was high in patients with anorectal malformations (ARM). The first significant attempt was made in 1783 to save these patients by making inguinal colostomy, but was unsuccessful. Later on, with the advent of new management modalities and a better understanding of anatomy and nature of these malformations, the outcome started improving.[1]

The major detrimental factor that affected the prognosis was a much higher incidence of associated anomalies with ARM. About 20-80% of patients with ARM have one or multiple associated anomalies. Complex urogenital, cardiac, and other system's anomalies had an adverse impact on the survival of such patients.[1,2]

Other factors such as low birth weight, delay in access to paediatric surgeon, septicaemia, gut perforation in combination with associated anomalies have an adverse impact on the prognosis.[3]

The mortality and morbidity rates are higher in patients with ARM who present in neonatal life, as compared to those in the infantile or older age. The underlying reason is the additional risk factors of neonatal life such as hypothermia, hypoglycaemia, poor immunity and sepsis.[3-6]

This prospective study was designed to evaluate the variable "associated congenital anomalies" as a factor of poor prognosis (mortality).

MATERIALS AND METHODS

This was a prospective observational study with statistical support, conducted at the Department of Pediatric Surgery, The Children's Hospital, and The Institute of Child Health, Lahore Pakistan. The study period was from March 2008 to January 2009. The sampling technique was purposive non-probability sampling. A structured proforma was developed for recording the demographic information, type of malformation, associated anomalies, management performed, and outcome of 100 consecutive patients

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presented with ARM in neonatal life.

After stabilising the patients in a neonatal surgical emergency, each patient was thoroughly examined, clinically, to identify the type of malformation, visible associated anomaly, any murmur, or other abnormalities. The Krickenbeck classification system was used to categorise these malformations. An invertogram was performed in patients with imperforate anus without clinically identified fistula to delineate the level of distal gas shadow. Ultrasound was performed in each patient to rule out urogenital anomalies. Echocardiogram was requested in patients with positive findings during auscultation of the chest.

Patients were categorized into two groups, based on the presence and absence of associated congenital anomalies.

A sigmoid loop colostomy over the skin bridge was performed for patients with recto-urethral/vesical fistula, imperforate anus without fistula having rectal gas shadow >1 cm from anal pit, recto-vaginal fistula, persistent cloaca, vestibular fistula where abdomen was distended due to stenotic opening, H-fistula, and rectal atresia. At the time of colostomy formation, the distal segment of colon was emptied and washed with normal saline to prevent urinary tract infections and faecaloma. Anoplasty was performed in perineal fistula, anal stenosis, and imperforate anus without fistula where rectal gas level was <1 cm from anal pit. Colostomy and anoplasty were performed under general anaesthesia in most patients; however, in patients who were sick and not fit for general anaesthesia, these procedures were performed under local anaesthesia. Exploratory laparotomy was performed in patients presenting with pneumoperitoneum, malrotation, congenital pouch, colon, etc. Dilatation of stenotic vestibular fistula was performed in patients with vestibular fistula not fit for general anaesthesia. Parents of patients with cloacal exstrophy were counseled about the complex nature of the malformation and advised them to visit after few months to be considered for staged correction of anomalies.

All clinical events were recorded and analysed using *EpiInfo* version 3.5.1, at the completion of 100 cases. The association of outcome and congenital anomalies was judged by Chi square test. A *P*-value of <0.05 was considered statistically significant to rule out the null hypothesis.

RESULTS

Of 100 patients with ARM, 77% were male and 23% were female (3.35:1). Age of presentation ranged from 12 hrs and 28 days (mean, 3.4 days). The mean weight of neonates presenting with ARM was 2.52 kg, with a range of 1.53.9 kg.

Major groups of these malformations, as per Krickenbeck classification system, were imperforate anus without clinically identified fistula (60%), recto-urethral/vesical fistula (13%), and vestibular fistula (8%). Table 1 shows relative frequencies of various types of ARM in either gender.

Twenty-eight percent of patients with ARM had associated congenital anomalies and syndromes. Of 28 patients, 10 had multiple associated congenital anomalies. The major groups of systems involved were urogenital in 10%, cardiovascular in 8%, and gastrointestinal in 6%. Down's syndrome was present in 8% patients. In one patient, OEIS (omphalocele, exstrophy, imperforate anus, and spinal defects) complex was present. Table 2 shows relative frequencies of associated anomalies encountered in neonates with ARM.

Table 1: Relative frequencies of types of anorectal malformations in either gender

Male		Female	
ARM	Frequency %	ARM	Frequency %
Perineal fistula	5	Perineal fistula	2
Recto-urethral/ recto-vesical fistula	13	Vestibular fistula	8
	54	Persistent cloaca	3
Without fistula	2	Without fistula	6
Congenital pouch colon	1	Recto-vaginal fistula	1
Rectal atresia	2	H-type fistula	1
Complex defects		Complex defects	2

ARM: Anorectal malformations. These percentages are taken from out of total patients (n=100). The term recto-urethral/recto-vesical fistula is used in combination due to inability to clinically differentiate between the two. Complex defects (cloacal exstrophy, OEIS complex)

Table 2: Relative frequencies of associated anomalies and syndromes

Associated anomalies/syndromes	Frequency %
Congenital heart diseases	8
Posterior urethral valve	3
Tracheo-oesophageal fistula	3
Limb anomalies	3
Cranio-facial anomalies	4
Malrotation	1
Hypospadias	1
Penile agenesis	1
Renal agenesis	1
Bladder exstrophy	3
Vaginal prolapse	1
Sigmoid colonic atresia	1
Down's syndrome	8
OEIS complex	1

These percentages are taken from out of total patients (n=100)

Sigmoid loop colostomy was performed in 66% of patients, anoplasty in 18% patients, and exploratory laparotomy in 8% patients. About 4% patients were managed by dilatation of stenosed vestibular fistula, while 3% of patients with complex malformation were counseled and surgical repair was deferred for few months.

Postoperative complications were identified in 15% of cases. Among complications, wound infection was encountered in 10 patients, pericolostomy evisceration of the intestine in 2 patients, and in one patient each, there was colostomy retraction and gangrene of colostomy. Four patients had a reoperation for these reasons.

The mortality rate in our series was 15%. Of 28 patients, 11 with associated congenital anomalies died. In contrast, 4 of 72, in the group without associated congenital anomalies, expired, generating a statistically significant difference in both groups ($P<0.05$). Table 3 describes the mortality with reference to associated anomalies and Table 4 describes the proportionate mortality as to the individual risk factor.

DISCUSSION

ARM are presumed to occur for centuries; however, limited prevenient record of such malformations may indicate high mortality associated with these malformations.[1]

Table 3: Mortality with reference to risk factors

Patients	Risk factors
A	TEF, Down's syndrome, limb anomalies
B	TEF, Severe pneumonia
C	CHD, LBW
D	Left renal agenesis, penile agenesis, CHD, sepsis
E	Down syndrome, CHD
F	Down syndrome, CHD
G	Down syndrome, CHD
H	OEIS complex
I	CHD, PUV, metabolic acidosis, sepsis
J	Pneumoperitoneum, no anomalies
K	Pneumoperitoneum, no anomalies
L	CHD (Tricuspid atresia)
M	TEF, LBW, sepsis
N	No anomalies
O	No anomalies

TEF = Tracheo-oesophageal fistula, CHD = Congenital heart disease, PUV = Posterior urethral valve

Table 4: Proportionate mortality with reference to risk factors

Risk factor	% mortality
TEF	20
CHD	46.7
Down's syndrome	13.33
Complex renal anomalies	26.67
Pneumoperitoneum	13.33
LBW	13.33
Sepsis	20

Multiple risk factors were present in most of patients (expired)

ARM is unique in many aspects and the most important is its higher association with congenital anomalies. The reported incidence of associated anomalies varies across the world in frequencies as well as type of associated anomalies. The reported range of associated anomalies varies between 20% and 80%. In our series, 28% of patients had associated anomalies that lie within the reported range.[2,4]

The most common associated anomalies reported from across the world, were related to the urogenital system. Other systems affected with associated anomalies were cardiovascular, gastrointestinal tract (GIT), vertebra, and nervous system. The most common urogenital anomalies associated with patients of ARM were renal agenesis, ectopic kidney, vesicoureteral reflux, hypospadias, and undescended testes. In our series, the system mostly affected was urogenital system and the associated anomalies were renal agenesis, hypospadias, posterior urethral valves, and bladder exstrophy. Some centres have built their own protocols to identify urogenital anomalies by thoroughly investigating the patient with all available imaging modalities. However, in our series, ultrasound of abdomen and clinical examination were used for detecting these anomalies in neonatal life. More sophisticated modalities, such as IVP, MCUG, CT scan, and cystoscopy, are usually used in the follow-up period.[7-11]

In our series, we encountered more patients without a clinically identified recto-urethral or recto-vesical fistula, in contrast with other series.[1] The proportion of these fistulae may change later on when a loopogram would be performed so as to delineate any fistulous communication with the urinary system. We labeled the ARM with a recto-urethral or recto-vesical fistula on the basis of clinical evidence of passing meconium with urine.

The mortality encountered in patients with ARM was high in the past due to lack of understanding of the nature and anatomy of these malformations along with problems of associated anomalies and risk of anaesthesia. As a result of advancements in diagnostic, therapeutic modalities, and development of better intensive care units, the mortality rate is reduced and the range now reported lies between 1.4% and 30%.[4-6,12]

The mortality rate in our series was 15%. Of the total

mortality rate, 73% was in the group with associated congenital anomalies and 26.6% in the group without associated congenital anomalies. The mortality was high in the group having associated congenital anomalies, despite the fact that the total number in this group was less than one-third of the total patients ($P<0.05$).

In concluding, mortality rate is higher in neonates with ARM having associated congenital anomalies. Cardiovascular anomalies, tracheo-oesophageal fistula, complex urogenital anomalies, pneumoperitoneum, sepsis, and low birth weight are risk factors and indicators of poor prognosis in such patients.

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