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Frequency of congenital cardiac anomalies in patients with anorectal malformations.

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ABSTRACT… Objectives: To determine the frequency of congenital cardiac anomalies in patients with anorectal malformations. **Study Design:** Retrospective study. **Setting:** Department of Pediatric Surgery, Khyber Teaching Hospital, Peshawar. **Period:** Jan 2018 to June 2018. **Material & Methods:** All patients from 0 to 30 days of life both males and females (Inclusion criteria) with diagnosed anorectal malformation on clinical and radiological assessment, underwent cardiovascular work up to identify any cardiovascular disease. Patients with intersex disorders and those operated elsewhere were excluded. Mean and standard deviation were calculated for age. Frequency and percentage were calculated for gender, type of anorectal malformation, genital ambiguity, previous surgical intervention and congenital cardiac anomalies. Data was stratified for age, gender, lesion level, genital ambiguity and previous surgery to see the effect modification. **Results:** Out of 90 patients with anorectal malformation, there were 49 (54%) males and 41 (46%) females (M: F;1:1.19). with mean age of 7.65 ± 2.43 days. There were 39 (43.34%) patients with low anorectal malformation and 51 (56.67%) patients with high anorectal malformation. Genital ambiguity and previous surgical intervention was present in 8 (8.89%) and 11 (12.23%) patients, respectively. Congenital Cardiac Anomalies were diagnosed in 16 (17.78%) patients with anorectal malformation. Congenital Cardiac Anomalies were mostly seen in age group 1-10 days (21.95%), male children (22.45%) and high anorectal malformation (23. 52%). **Conclusion:** The frequency of congenital cardiac anomalies in patients with anorectal malformations was found lower than local figures. The majority of congenital cardiac anomalies associated with high anorectal malformation and male gender.

Key words: Anorectal Malformations, Congenital Cardiac Anomalies, Genital Ambiguity, Gastrointestinal Anomalies, High Anorectal Malformations.

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INTRODUCTION

The co-existence of congenital heart diseases (CHD) with gastrointestinal (GI) tract anomalies is not an uncommon finding. So far, the reported association of these anomalies are highly variable. Anorectal malformations (ARMs) occupy a major bulk of congenital anomalies commonly encountered in Paediatric surgery, with an estimated incidence ranging between 1 in 2000 and 1 in 5000 live births. The condition comprises a broad spectrum of defects ranging from slightly abnormal positioned anus with excellent functional outcomes to complicated anatomical deviation of the gut and urogenital system where management becomes challenging.[1,2]

ARMs are not uncommon among anomalies of neonates. This embryological and fetal mishap happens as a result of antenatal maldevelopment of cloaca and urorectum. Recently survival rate of ARMs has significantly evolved because of progress in surgical techniques, better understanding and availability of improved neonatal surgical and intensive care facilities. Mostly ARMs are diagnosed after birth, and co-exist with other anomalies; mostly, urological, cardiac, and musculoskeletal. Fistulous communication with urethra in males and vestibule in females are the most common variants. ARM represent a special class of the anomalies of anus and rectum unique in a way that it has higher association with other birth disorders. The reported range

of associated anomalies varies from 20% to 80%.[3] The most common associated anomalies reported from across the world, are urogenital in origin. Other systems affected with associated anomalies are cardiovascular, gastrointestinal tract (GIT), vertebra, and nervous system.[4,5]

Unlike normal newborns, the frequency of CHDs is greater in patients with malformations of GIT. In general population the incidence of CHD is <1%, but the significant association between major GI malformations and CHDs has been historically emphasized.[6,7]

Previously, a study by Gokhroo RK et al, found anorectal malformation (74.41%) as the most common birth defect of gastrointestinal tract, of which 50% h ad congenital heart disease.[8] While in another study by Kamal JS, et al, the frequency of congenital cardiac abnormalities among the imperforate anus subjects was 24.6%.[9] In a retrospective study by Schierz IAM et al, the frequency of congenital heart disease in newborns with apparently isolated single gastrointestinal malformation was 15.5%.[10] The rates of cardiac malformations shown in these studies is highly variable with no close consensus.

Case series by Qazi SH et al, in Karachi reported that the prevalence of cardiac abnormalities in subjects with ARMs was 38% while a prospective observational study in Lahore, reported that the incidence of cardiac abnormalities was 8% in patients with anorectal malformation.[11,12]

In literature, various authors have described a variable frequency of congenital heart diseases among patients with ARM. In study by Schierz IAM et al, the frequency of CHD among patients with ARM was as low as 15.5% whereas, Gokhroo RK et al, described as high as 74.4%. Even our local data shows variability among different authors ranging from 8 – 38%. So, there was a need to conduct more studies to know the frequency of CHD among patients with ARM from a sample representing our population.

I wanted to conduct this study with the objective that the frequency of research will depict the burden of cardiac care needed for these patients. Moreover, the, results of the study might catch the attention of cardiac surgeons in Pakistan as it would be a local data and the approach to patients with both ARMs and CHDs may become more multi-disciplinary.

MATERIAL & METHODS

After formal ethical approval, retrospective study included 90 patients (both male and female) with anorectal malformation and congenital cardiac anomaly of any variety admitted to paediatric surgical unit Khyber teaching hospital Peshawar between June 2018 to June 2019. Age of patients ranged from 0 to 30 days. Neonates with genital ambiguity and those who had undergone any operation somewhere else were excluded. Patients with diagnosed anorectal malformation on clinical and radiological assessment underwent cardiovascular work up i.e. physical examination, ECG, CXR and Echocardiography to identify any cardiovascular disease and were labeled as 'Yes" for congenital cardiac anomaly (as per operational definition). All the information was recorded on printed proforma (attached). Confounders in the study were controlled by strictly obeying the pre decided criteria to include or exclude subjects. Data was entered and analyzed in SPSS version 20.0 computer program. Mean and standard deviation were calculated for quantitative variables like age. For qualitative variables like gender, type of anorectal malformation, genital ambiguity, previous surgical intervention and congenital cardiac anomalies frequency and percentage were calculated. Data was stratified for age, gender, lesion level, genital ambiguity and previous surgery elsewhere to see the effect modification. Chi square test was applied after stratification and P –value ≤0.05 was assumed as significant.

RESULTS

There were 23 (25.56%) patients with anorectal malformation in the age group < 1 day, 41 (45.56%) patients in the age group 1 – 10 days, 19 (21.12%) patients of age 11 – 20 days and 7 (7.76%) patients in the age group 21 – 30 days. The mean age of the patients with anorectal malformation was 7.65 ± 2.43 days. (Table-I).

Regarding gender distribution, out of 90 patients with anorectal malformation included in study, there were 49 (54%) males and 41 (46%) females with M: F ratio of 1:1.19. Among these there were 39 (43.34%) patients with low anorectal malformation, 51 (56.67%) patients with high anorectal malformation. (Table-II). 8 (8.89%) patients had genital ambiguity and genital ambiguity was not seen in 82 (81.12%) patients. (Table-III). In our study, there were 11 (12.23%) patients with previous surgical intervention history and previous surgical intervention was not done in 79 (87.78%) patients. (Table-IV)

Out of 90 patients with anorectal malformation included in our study, there were 16 (17.78%) patients with congenital cardiac anomalies and congenital cardiac anomalies was not seen in 74 (82.23%) patients. (Table-V)

Stratification of data (Congenital Cardiac Anomalies) with effect modifier (Age): Out of 23 patients with anorectal malformation, in age group of <1 day, 4 (17.39%) patients showed Congenital Cardiac Anomalies and in 19 (82.60%) patients, no Congenital Cardiac Anomalies was diagnosed. Out of 41 patients with anorectal malformation, in age group of 1–10 days, 9 (21.95%) patients showed Congenital Cardiac Anomalies and in 32 (78.04%) patients, no Congenital Cardiac Anomalies was diagnosed. Out of 19 patients with anorectal malformation, in age group of 11– 20 days, 2 (10.52%) patients showed Congenital Cardiac Anomalies and in 17 (89.47%) patients, no Congenital Cardiac Anomalies was diagnosed. Out of 7 patients with anorectal malformation, in age group of 21 – 30 days, 1 (14.28%) patient showed Congenital Cardiac Anomalies and in 6 (85.71%) patients, no Congenital Cardiac Anomalies was diagnosed. The p-value was 0.719. (Table-VI)

Stratification of data (Congenital Cardiac Anomalies) with effect modifier (Gender): Out of 49 male patients with anorectal malformation, 11 (22.45%) patients showed Congenital Cardiac Anomalies and in 38 (77.55%) male patients, no Congenital Cardiac Anomalies was diagnosed. Out of 41 female patients with anorectal malformation, 5 (12.19%) patients showed Congenital Cardiac Anomalies and in 36 (87.80%) female patients, no Congenital Cardiac Anomalies was diagnosed. The p-value was 0.0592. (Table-VII)

Stratification of data (Congenital Cardiac Anomalies) with effect modifier (Anorectal malformation level): Out of 39 patients with low anorectal malformations, 4 (10.25%) patients diagnosed with Congenital Cardiac Anomalies and no Congenital Cardiac Anomaly was seen in 35 (89.74%) patients. Out of 51 patients with high anorectal malformations, 12 (23.52%) patients diagnosed with Congenital Cardiac Anomalies and no Congenital Cardiac Anomaly was seen in 39 (76.47%) patients. The p-value was 0.0691. (Table-VIII)

Stratification of data (Congenital Cardiac Anomalies) with effect modifier (Genital ambiguity): Out of 8 patients with Genital ambiguity, 1 (12.5%) patient showed Congenital Cardiac Anomaly and 7 (87.5%) patients did not have any congenital cardiac anomalies. The p-value was 0.820. (Table-IX)

Stratification of data (Congenital Cardiac Anomalies) with effect modifier (Previous Surgery): Out of 11 patients with history of Previous Surgical intervention, 2 (18.18%) patient showed Congenital Cardiac Anomaly and 9 (81.81%) patients did not have any Congenital Cardiac Anomaly. The p-value was 0.647. (Table-X)

Age (Days)	No. of Patients (n)	Percentage (%)
<1	23	25.56
1-10	41	45.56
11-20	19	21.12
21-30	7	7.76
Mean±SD	7.65 ± 2.43 days	

Table-I. Distribution of patients by age (n=90)

Types of Anorectal Malformation	Number of Patients	
	Frequency	Percentage (%)
Low	39	43.34
High	51	56.67
Others	0	0

Table-II. Distribution of patients by types of anorectal malformation (n=90)

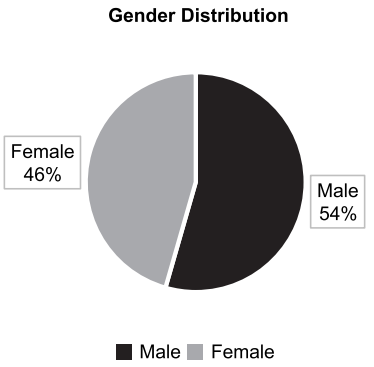

Figure-1. Distribution of patients by gender (n=90)

Genital Ambiguity	Number of Patients (n=90)	
	Frequency	Percentage (%)
Yes	8	8.89
No	82	81.12

Table-III. Distribution of patients by genital ambiguity

Previous Surgical Intervention	Number of Patients (n=90)	
	Frequency	Percentage (%)
Yes	11	12.23
No	79	87.78

Table-IV. Distribution of patient by previous surgical intervention

Congenital Cardiac Anomalies	Number of Patients (n=90)	
	Frequency	Percentage (%)
Yes	16	17.78
No	74	82.23

Table-V. Distribution of patient by congenital cardiac anomalies

Age Groups (days)	Congenital Cardiac Anomalies			
	Yes		No	
	No.	%	No.	%
<1 (n=23)	4	17.39	19	82.60
1-10 (n=41)	9	21.95	32	78.04
11-20 (n=19)	2	10.52	17	89.47
21-30 (n=7)	1	14.28	6	85.71
P value*	**0.719			

Table-VI. Stratification of data (Congenital Cardiac Anomalies) by effect modifier (age) (n=90) *Chi square test, *Not Significant

Gender	Congenital Cardiac Anomalies			
	Yes		No	
	No.	%	No.	%
Male (n=49)	11	22.45	38	77.55
Female (n=41)	5	12.19	36	87.80
P value*	**0.0592			

Table-VII. Stratification of data (Congenital Cardiac Anomalies) by effect modifier (Gender) (n=90) *chi square test, **Not Significant

Anorectal Malformation Level	Congenital Cardiac Anomalies			
	Yes		No	
	No.	%	No.	%
Low (n=39)	4	10.25	35	89.74
High (n=51)	12	23.52	39	76.47
Others	0	0	0	0
P value*	**0.0691			

Table-VIII. Stratification of data (Congenital Cardiac Anomalies) by effect modifier (anorectal malformation level) (n=90) *chi square test, **Not Significant

Genital Ambiguity	Congenital Cardiac Anomalies			
	Yes		No	
	No.	%	No.	%
Yes (n=8)	1	12.5	7	87.5
P value*	**0.820			

Table-IX. Stratification of data (Congenital Cardiac Anomalies) by effect modifier (Genital Ambiguity) (n=90) *chi square test, **Not Significant

Previous Surgery	Congenital Cardiac Anomalies			
	Yes		No	
	No.	%	No.	%
Yes (n=11)	2	18.18	9	81.81
P value*	**0.647			

Table-X. Stratification of data (Congenital Cardiac Anomalies) by effect modifier (Previous Surgery) (n=90) *chi square test, **Not Significant

DISCUSSION

Association between GI(gastrointestinal) malformations and congenital heart diseases is well established. Objective of the study was to know the frequency of congenital cardiac anomalies occurring in association with anorectal malformations.

The mean age of the neonates with anorectal

malformation was 7.65 ± 2.43 days in our study. In a study by Qazi SH et al, the median age at presentation was the 1 day of life, as 56% of neonates were brought immediately after birth.[11] Among 90 patients with anorectal malformation included in study, 49 (54%) males and 41 (46%) females with M: F ratio of 1:1.19. In a study by Qazi SH et al, in sample of 84 68 % were males whereas 32% were females.[11] In a study by Kamal JS and Azhar AS, the male to female ratio was 32:29.[9] In a study by Gokhroo RK et al, among 43 patients, 79% were males and 21% females.[8] In a study by Örün UA et al, males were 55.8% and females 44.2 %, with their ages ranging from 0 to 15 years.[14] In a study by Teixeira 0HP et al, there were 43 boys and 25 girls with imperforate anus.[13]

There were 39 (43.34%) patients with low anorectal malformation, 51 (56.67%) patients with high anorectal malformation in our study. In a study by Teixeira 0HP et al, 12 patients had type I anorectal anomaly, 1 had type II, and the remaining had type III.[13] Out of 90 patients with anorectal malformation that included in our study, there were 8 (8.89%) patients with genital ambiguity and there were 11 (12.23%) patients with previous surgical intervention history.

In our study, there were 16 (17.78%) patients with anorectal malformation who were diagnosed with congenital cardiac anomalies. In a study by Qazi SH et al, Cardiac anomalies were the leading associated anomalies with anorectal malformations and urological anomalies in second most frequent.[11] In a study by Kamal JS and Azhar AS, the rate of congenital cardiac anomaly among anorectal malformation subjects was 15 (24.6%), that is, nine males and six females.[9] In a study by Schierz IA et 15.5% of patients had concurrent CHDs.[10] Gokhroo RK et al, showed 60.46% patients had congenital heart diseases.[8] Study by Tulloh et al, presented that about 20% of patients with major GI malformations had an associated CHDs.[15] work by Chéhab et al depicted congenital cardiac anomalies in 38% of 105 patients with GI malformations.[16] Olgun et al showed the rate of CHD in patients with imperforate anus, as 15.9%, 28.6%, 23.7%.[17] Thompson et al reported different rates of CHD , in the same patient groups.[18] In a study by Örün UA et al, Congenital heart defects were recorded in 28.5% newborns with no statistically considerable difference between genders ($p>0.05$). Among 227 patients with a single gastrointestinal system malformation,26.8% cases were with congenital heart diseases; and, of 15 patients with more than one gastrointestinal system malformation, 53.3% had congenital heart disease ($p<0.05$).[14] In a study by Teixeira 0HP et al, 15 patients were with both cardiovascular anomalies and imperforate anus.[13]

We cross tabulated the congenital cardiac anomalies with age and found that the maximum frequency of congenital cardiac anomalies i.e. 21.95% were noted in age group 1-10 days, followed by < 1-day age (17.39%) 21-30 days (14.28%) and 11-20 days (10.52%). But this was not statistically significant. We also cross tabulated the congenital cardiac anomalies with gender and found that the maximum frequency of congenital cardiac anomalies i.e. 22.45% was noted in male gender as compared to female gender i.e. 12.19% however, it was not statistically significant.

We cross tabulated the congenital cardiac anomalies with levels of anorectal malformation and found that the congenital cardiac anomalies were more common in high level anorectal malformation i.e. 23.52% as compared to low anorectal malformation i.e.10.25%. But this was not statistically significant. The study has certain limitations. It was carried in single center on limited population size.

CONCLUSION

From the results of present study, we found that the frequency of congenital cardiac anomalies in patients with anorectal malformations lower than local figures. The majority of congenital cardiac anomalies associated with high anorectal malformation and male gender. But, this was not statistically significant. We need more studies on larger population size in multiple centers to verify this pattern before making any recommendation.

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AUTHORSHIP AND CONTRIBUTION DECLARATION

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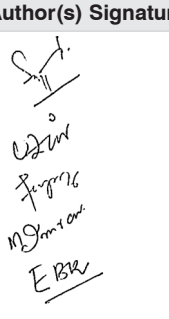
1	Sajjad Ali	Conception, Interpretation of data, Drafting, control review & final approval of version to be published.	
2	Muhammad Uzair	Study design & data analysis & interpretation of draft.	
3	Fayaz ur Rehman	Drafting manuscript & criticaly review.	
4	Mohammad Imran	Acquisition of data drafting manuscript & final approval.	
5	Erum Behroz Khan	Analysis of data & critically review.	
6	Muhammad Asim Khan	Interpretation of data & Final approval.	