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Five Year Experience of Anorectal Malformation with Esophageal Atresia of Tertiary Care Hospital

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Abstract: ***Five year experience of Anorectal Malformation with Esophageal atresia of Tertiary care hospital. Aim: We performed this study with an aim to compare all cases Esophageal atresia (EA+/-fistulae) with anorectal malformation (ARM) and only ARM, in terms of incidence, sex predilection, birth weight, surgical approach, outcome & mortality. Material & Method: Retrospective review of cases with Clinical data (from April 2012 - April 2017). The subjects of this study were 236 patients who had been diagnosed and managed for ARM. Among these patients, 25 patients associated with esophageal atresia were selected as the subject patient group. Result: Incidence of TEF WITH ARM was 11.1%. Study has more male preponderence. All cases are of Type c except 2 cases of type a. According to the classifications of anorectal malformations, there were 2 cases with rectourethral fistula and 8 cases with rectoperineal fistula and covered anus in the males. In the females, there was a varied distribution of 7 cases. There were 1 cases (4%) presenting as a part of the VACTERL association, which is the representative example of a complex anomaly. Most of cases died due to cardiac problem and pnemonitis (due to delayed presentation). Conclusion: The study concludes the experience of EA (+/-fistulae) with ARM, their distribution, incidence and outcome of tertiary care centre.***

Keywords: Anorectal Malformation with esophageal atresia, Tracheoesophageal fistulae, VACTERL (Vertebrae, anorectal malformation, cardiac, tracheoesophageal fistulae, renal anomalies, limb anomalies), rectoperineal fistulae

1. Introduction

Anorectal malformations (ARM) occur approximately in 1/1, 500 to 1/5, 000 live births [1, 2]. Commonly, we see either ARM or Tracheoesophageal fistulae but sometime we get both which is surgically challenged case Type of management may be staged or single stage depending on other associated anomalies and our anaesthesioa or post operative critical care facilities.

The frequency of other associated anomalies is approximately 40-70% in the decreasing order of urogenital system, musculoskeletal system, and cardiovascular system [3, 4]. In addition, anomaly in other parts of the gastrointestinal system can occur concomitantly, and immediate surgery required preventing complications.

Previously some studies were done and we are conducting the present study to analyze the characteristics of and treatment results in neonates with anorectal malformations associated with esophageal atresia at our tertiary centre.

2. Materials and Methods

Retrospective review of cases with Clinical data (from APRIL 2012 - APRIL 2017).

The subjects of this study were 236 patients who had been diagnosed and managed for anorectal malformations in the neonatal intensive care unit, Paediatric surgery department of SS hospital, BHU Varanasi, from April 2012- April 2017. Among these patients, 25 patients associated with esophageal atresia were selected as the subject patient group.

3. Methods

A retrospective study was conducted by using the clinical data of the 236 patients. First, clinical aspects such as the type of anorectal malformations and the frequency of other associated anomalies were compared between the groups with and without esophageal atresia. Comparative analysis of the clinical characteristics of anorectal malformations associated with esophageal atresia, as well as the processes of management and the methods, and results for the treatment in neonatal period, were performed.

4. Results

Comparison of groups with and without esophageal atresia

Among the 236 patients with anorectal malformations, there were 25 patients with esophageal atresia and 211 patients without it; the male to female ratio was 2.5: 1 (18:7) and 1.18: 1 (109:92), respectively. Regarding the type of anorectal malformations, there were 10and 100 cases of low-type malformation, which is correctable through a single procedure, in the group with and without associated esophageal atresia. There were 13 and 40 cases of high-type malformations with esophageal atresia or without atresia that is managed by staged or single stage Abdominoperineal pull through operation respectively. There were 2 and 41 cases of Intermediate Anorectal malformations with and without atresia. Cloacal anomaly was identified in 14 Anorectal malformation without esophageal atresia. (table 1).

Table 1: Clinical Aspects according to Accompanying Esophageal Atresia

	ARM WITH TEF(n=25)	ARM(n=211)
Sex(M:F)	18:7	119:92
High ARM	13	40
Intermediate ARM	2	14+27
Low ARM	10	65+51
Cloaca	0	14

Clinical characteristics of anorectal malformations associated with esophageal atresia

There were 25 neonates with esophageal atresia, accounting for 11.1% of the total subjects. Of them, male was 18 and female was 7; there was more preponderence in male in occurrence. According to the classifications of anorectal malformations, there were 2 cases with rectourethral fistula

and 8 case with rectoperineal fistula and covered anus in the males. In the females, there was a varied distribution of 7 cases in, 2 cases in rectovaginal fistula, and 1 case in rectovestibular fistula and 4 cases of Anovestibular fistulae (low type). In cases of esophageal atresia, there were 3 cases of type A esophageal atresia without tracheoesophageal fistula, but most cases were type C esophageal atresia with tracheoesophageal fistula (Table 2).

Table 2: Demographic and Clinical Characteristics of Anorectal Malformations Associated with Esophageal Atresia

Case	Sex	GA(wk)/BW(gm)	Type of ARM	Type of esophageal atresia
1	M	38/2300	H	C
2	F	28/1000	AVF	C
3	F	37/2200	AVF	C
4	M	34/1400	H	A
5	M	30/1300	H	C
6	M	40/2300	H	C
7	M	37/2400	L	C
8	M	38/2500	L	C
9	F	37/2100	AVF	C
10	M	33/2000	I	C
11	M	34/1800	H	A
12	M	33/1800	H	C
13	M	40/2500	I	C
14	M	39/3000	H	C
15	F	39/2000	L	C
16	F	36/2350	RECTOVAGINAL	C
17	M	37/2500	L	C
18	F	38/2300	POUCH	C
19	M	37/2300	I	C
20	M	37/2400	L	C
21	M	37/2200	H	C
22	M	34/2300	L	C
23	F	36/2300	POUCH	C
24	M	40/2500	H	C
25	M	38/2300	L	A

Distribution of associated anomalies

Associated anomalies in other organ system were confirmed in all the study cases and more than 3 associated anomalies were observed especially in 10 cases. When considering the frequency by organ system, there were 6 cases involving the cardiovascular system; 3 duodenal atresia; 1 malrotation, 2 meckels diverticulum; 1ileal atresia. In particular, there were 1 cases (4%) presenting as a part of the VACTERL (Vertebral abnormalities, Anal atresia, Cardiac anomalies, Tracheoesophageal fistula, Esophageal atresia; Renal and Limb anomalies) association, which is the representative example of a complex anomaly. There were 2 cases of Down syndrome. However, in anorectal malformations without esophageal atresia, there was a relatively low frequency of associated anomalies (23%), as compared to that in the study (64%) subject group; on the other hand, there were a few cases of autosomal abnormalities (Table 3).

Table 3: Comparison of Associated Anomalies between Two Types

Organ system	ARM WITH TEF	ARM
CVS	6	15
DA	3	8
Malrotation	1	1
Meckles diverticulum	2	2
Down syndrome	2	5
Ileal atresia	1	2
VACTERAL	1	2

Managements and results in the neonatal period

In 5 cases, a primary correction was done, 3 cases primary repair and Abdominoperineal pull through and 2 cases undergone primary repair and diverting colostomy. In 6 cases, anoplasty and a primary correction for esophageal atresia were performed concurrently. Diverting colostomy was performed first in 5 cases; one case had a delayed diagnosis of esophageal atresia and the other case had concomitant perforation in the gastrointestinal tract.2 cases undergone primary repair for atresia after PSARP.Remaining 3 of Anovestibular with atresia undergone primary repair and delayed repair for anorectalmalformation. The remaining three patients were confirmed to be dead due to associated anomalies in the other organs, without having received other surgical treatments (Table 4).

Table 4: Management & Clinical outcome

Method of management	Result(mortality)
Primary repair+ colostomy (2)	0
Primary repair after colostomy(5)	1
Primary repair after PSARP (2)	0
Primary repair +APPT(3)	1
Primary repair +anoplasty(6)	0
Primary repair +Anal dilation for AVF(4)	1
No treatment(3)	3(pre operative)

Management and Clinical Results in Neonatal Periods

There were 6 mortality cases in 25 patients (mortality rate, 24%).3 cases died preoperative without undergoing surgery. A more detailed observation showed that 3 case, a previously described case of death without surgical interventions, in which a female neonate with VACTERL association who was born at 28 weeks of gestation with a birth weight of 1, 000 g and rest 2 comes after 7 days with respiratory failure,.

In the other 2 cases, the cause of death was a heart problem due to associated cardiac anomalies, and 1 of these cases was confirmed to be a male neonate in whom a diagnosis of esophageal atresia was delayed because of a perforation in the gastrointestinal tract. Overall, for the 22 cases with receiving a correction surgery in the neonatal period, 3 cases of death were observed in the postoperative period. The cause of death in these cases was confirmed to be attributable to other associated anomalies rather than the disease of focus in the present study. Meanwhile, in the subjects with anorectal malformations without esophageal atresia, 5 case of death was observed (mortality rate, 2.33 %). This was not a case of neonatal period, but due to a heart problem developed in follow-up period after final operation. (Table 4)

5. Discussion

The most frequently associated anomalies with EA±TEF are cardiac (49%) and anorectal malformations (15%). [5-8] Previous various articles concluded esophageal atresia in patients with anorectal malformations may occur alone or as a part of complex anomaly, and its incidence is reported

approximately in 8-11% [3, 9] The incidence rate in the present study was 11.1%, which was not largely different from that in other reports. Moreover, associated anomalies other than anorectal malformations and esophageal atresia were observed in all patient subjects. The majority of these anomalies were problems of the cardiovascular or urogenital systems, and they were not largely different from those reported in previous studies [10].

However, a relatively higher frequency of anomalies was observed in the subjects with esophageal atresia than in those without esophageal atresia. Thus, given that additional associated anomalies were observed in all patients, it is suggested that the management of anorectal malformations associated with esophageal atresia will require more caution and efforts than the management of patients presenting with anorectal malformations alone. Although many factors may be involved in the occurrence of these anomalies, it may be thought that anorectal malformations and esophageal atresia are types of midline defects, which present along the body. These defects occur as results of the combination of deficits in mesodermal migration and endodermal defects [11, 12]

On the other hand, in VACTERL association, there are two different explanations involving it; genetic factors and external environmental factors. First, the genetic factor model has been supported through animal experiments, and it proposes the formation of anomalies due to genetic mutations that induce signalling pathway abnormalities [13-15].

Second, it is thought that external environmental factors such as maternal diabetes, hormonal exposure during infertility treatments, and exposure to toxic factors negatively influence the morphological development of the fetus, leading to malformations in the fetus [16-19]

In our study, we could not identify any factors, such as chromosomal defects, which may induce this anomaly, but it is necessary to be supposed to think about an adequate investigation of these factors. However, since the incidence of the disease itself is not very high, there are still many limitations to investigate directly.

Generally, associated anomalies occur more frequently in high-type of anorectal malformations than in low-type malformations; the frequency is also higher in males than in females [7, 22-23]. In our study, for anorectal malformations associated with esophageal atresia, the gender distribution was MORE COMMON IN Male, with a 2.25: 1 male to female ratio. In the case of male, most of anorectal malformations were LOW - or high-type with rectourethral fistula (6/18, 33.3%) and high7/18 (38.88%), and mainly low-type malformations in female (4/7)57.11%. These findings showed a different distribution in cases of anorectal malformations without esophageal atresia. A relatively high frequency, 28.6%, of the cases occurred as a part of the VACTERL association, which may be considered as result of multiple congenital malformations. In addition, considering the report that the frequency of concomitant gastrointestinal atresia is 4 times as high in VACTERL association than in the cases with a single anomaly of anorectal malformation, this can also be inferred as a result of simple primary malformation due to deficits in the organ formation process, in addition to the previously mentioned disease occurrence process [21-23].

In the management of a complex anomaly, the correlation with life support must be considered primarily in the neonatal period, and it is essential to do appropriately a staged procedure. Ultimately, when considering it's clinical results, the complicated nature of the surgical procedure and appropriate surgical approaches must be considered in managing a case of anorectal malformations associated with esophageal atresia. In this study, the majority of the surgical treatments administered in the neonatal period were a staged procedure with good results, and there were no major differences compared to other reports [24-26]. Although the mortality rates after appropriate treatments are relatively low in the cases of simple anorectal malformations and esophageal atresia, the total mortality rate observed in the present study was 24%, a very high frequency compared to that in other gastrointestinal anomalies. In particular, when compared to anorectal malformations without esophageal atresia, it can be concluded that this high mortality rate is attributable to complications associated with other anomalies, rather than the disease itself. Two of the 3 deaths (pre operative) in our study were caused by heart problems during observation after the primary surgery in the neonatal period, and 1 death in VACTERL associations without surgical intervention. Thus, it could be inferred that the influence of other associated anomalies was greater than that of anorectal malformations and esophageal atresia. Meanwhile diverting colostomy was performed initially in 1 patients of 3 died post operative. Each case involved a delayed diagnosis of esophageal atresia or concomitant gastrointestinal perforation. Because of these complications, complex surgical procedures were unavoidable, and one case involved with gastrointestinal perforation resulted in a death during the postoperative recovery period. This indicates that a more careful and timely diagnosis based on clinical and radiological findings is necessary before the primary operation.

6. Conclusion

Anorectal malformations associated with esophageal atresia, whether alone or as a part of a complex syndrome, had a relatively high frequency of associated anomalies in other organs, as well as a high mortality rate. It may be suggested that a staged and multidisciplinary approach is essential in the management of neonates with this anomaly. In addition, reasonable treatments considering these possibilities may lead to improved outcomes by preventing delays in the diagnosis of anorectal malformations associated with esophageal atresia, as well as the development of serious complications.

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