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FREQUENCY OF VERTEBRO-SPINAL ANOMALIES IN PATIENTS PRESENTING WITH ANORECTAL MALFORMATIONS. A DEVELOPING COUNTRY, SINGLE CENTRE STUDY

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ABSTRACT

Background: To determine the frequency of different vertebro-spinal anomalies in patients with anorectal malformation presenting in developing country.

Study Design: Prospective observational study

Duration of study: Duration of study is 6 months.

Methods: After approval from the institutional review board, 106 pediatric patients after fulfilling selection criteria were enrolled in the study using non-probability convenience sampling technique. Informed consent was obtained from the guardian of patients. Baseline data of enrolled patients were recorded after careful history taking and examination. Subsequently, these patients underwent imaging through whole body X-rays and spinal cord MRI (Magnetic resonance imaging). Findings in the MRI were noted. The collected data was analyzed through SPSS v.26.

Results: We enrolled 106 patients in current study with mean age was 4.48±2.84 years and birth weight was found 2.58±0.52 kg. Out of 106 enrolled patients 62(58.5%) were male and 44(41.5%) patients were female. The male to female ratio was 1.41:1 in current study. The frequency of vertebro-spinal anomalies were found in current study as: 8(7.5%) patients had sacral agenesis, 4(3.8%) patients had scoliosis, 6(5.7%) patients had tethered cord and 2(1.9%) patients had thickened

filum terminale. Overall frequency was 18.9%.
Conclusion: 18.9% of ARM (anorectal malformation) patients were found to have vertebro-spinal anomalies. Sacral agenesis was present in 7.5% of patients (n = 8) and Tethered cord was present in 5.7% of patients (n=6). The presence of VACTERL-association was shown to be associated to the presence of vertebrospinal anomalies. Therefore, we emphasize the importance of screening for vertebro-spinal anomalies in patients with ARM, regardless of ARM type.

Trial Registration:
The study is registered with Clinical Trials and registration number is NCT06234020.
Registration Date: 22/01/2024.

What is already known:

- The reported frequency of vertebrospinal anomalies in ARM patients varies, with studies showing rates between 20% to 50%, depending on the population and diagnostic methods used.
- Common vertebrospinal anomalies include sacral agenesis, hemivertebrae, scoliosis, tethered cord, and spina bifida.
- Higher association in complex ARM: Patients with high or complex ARM tend to have a higher prevalence of vertebrospinal anomalies compared to those with low ARM.

What this study adds:

- The frequency of vertebrospinal anomalies in ARM patients in developing country.
- Highlights differences in diagnosis, management, and outcomes compared to studies from developed countries.
- Investigates whether the frequency of vertebrospinal anomalies varies based on the type and complexity of ARM in this population.

INTRODUCTION:

Anorectal malformations (ARMs) occurs with incidence of 2 to 6 per 100,000 of the population [1] with average of 3.5 per 100,000 in England [2]. Factors associated with decreased survival rates are low birth weight and co-existence of ≥2 other anomalies. Highest 10-year survival rate i.e., 100% is observed in patients with isolated ARMs independent of their birth weight. Abnormal cloacal membrane formation, which is implicated in the pathophysiology of ARMs, leads to the disrupted development of muscular and nervous structures [3].

Genetic counseling in ARMs should be done as it had high incidence of syndrome forms. Associated mutations [4] can be de novo pathogenic frame shift variants in ADNP, variants in *BBS1, CREBBP, EP300, FANCC, KDM6A, SETD2*, and *SMARCA4* genes. Common syndromes in which ARMs are seen include Helsmoortel-van der Aa syndrome, VACTERL association, Bardet-Biedl syndrome, Fanconi anemia, Rubinstein-Taybi syndromes, Kabuki syndrome 2, and Coffin-Siris syndrome [5]. This necessitates the screening of these patients for other anomalies [6].

VACTERL associations have been found in 16-17% [7] of the patients screened for additional anomalies already diagnosed with ARMs. Associated anomalies are found to exist in decreasing order of prevalence [8] in genitourinary, cardiovascular, and vertebro-spinal systems (28%, 25% and 22.6% respectively).

Fanjul et al [9] studied gynecological anomalies and observed that 50.8% of the female patients with ARM have any form of müllerian abnormalities; 30.2% have any congenital vaginal anomaly; 48.4% have associated uterine malformation and 9.5% have tubal problems. Most frequent vaginal anomalies were the complete longitudinal vaginal septum and vaginal agenesis whereas

the commonest uterine anomalies were didelphys and septate uterus. VACTERL association [10] itself does not increase the risk of gynecological anomalies in such patients, but VACTERL with rectoperineal and rectovaginal fistulas correlates positively with the risk of gynecological anomalies in females with ARMs.

Isolated urological anomalies excluding neurogenic bladder due to spinal cord anomalies which coexist with ARM[11] are hydronephrosis, unilateral renal agenesis, renal dysplasia, and ectopic kidney; vesicoureteric reflux is observed in up to 5.1% of patients. Rarely, urethral duplication and occult spinal dysraphism can be seen with ARM [12].

Vertebro-spinal anomalies are not uncommon in patients with anorectal malformations, and can range from asymptomatic anomalies (e.g., ventriculus terminalis or filum cysts) to tethered cord symdome, spinal lipoma, lipomeningocele, neural tube defects and diastematomyelia [13] which require neurological interventions. Hence, pediatric patients diagnosed with ARM must be screened through spinal ultrasonography for relevant spinal cord anomalies [14]. Similarly cervical ribs (usually rudimentary), deviated number of thoracic ribs, and presence of lumbar ribs have been found in patients with ARM too [15].

Early identification is very important to identify vertebro-spinal anomalies in patients with ARM. Failure in identification these abnormalities could lead to future damage, such as neurogenic bladder disorder in patients with TSC, which may result in vesico-urethral reflux, or urinary incontinence. Hence, it is advisable to do screenings for vertebrospinal anomalies in all patients with ARM.[16]

MRI is widely regarded as the most accurate method for identifying spinal cord issues, ultrasonography[17] of the spinal cord can also be a reliable and faster alternative imaging technique that is more cost-effective. However, the visualization of the spinal cord via ultrasound becomes challenging in later stages of life due to the development of bone, particularly if the ultrasound is conducted after neonatal period.

ARM is very common among Pakistani population but association of other anomalies as well as their prevalence has not been studied in our population this far. Our study aims to conduct this study to find frequency of associated vertebro-spinal anomalies in pediatric patients with ARM.

MATERIALS And METHODS:

After approval from the institutional review board, 106 pediatric patients presenting in the Outpatient Department of Children Hospital, in the Department of Pediatric Surgery from January, 2024 to July, 2024 with diagnosis of ARM were enrolled in study using non-probability convenience sampling technique. Patient selection was made in the light of selection criteria.

Inclusion criteria includes:

Patients presenting in OPD with diagnosed cases of ARM.

Patients giving consent to participate in study.

Exclusion criteria includes:

1. Patients presenting in emergency.
2. Patients already admitted in ward.

All patient guardians were told about the research and asked for their informed permission. The recruited patients' baseline data was documented after a thorough history and examination. These patients then had spinal cord MRI and whole body X-rays were performed for investigative purpose. The results of the MRI were recorded.

STATISTICAL ANALYSIS:

The collected data was analyzed through Statistical Package for the Social Science version 26.0. Population characteristics which are categorical e.g., sex, type of ARM according to Krickenbeck classification, mode of delivery, frequency of vertebrospinal anomalies, birth weight category, and

presence of VACTERL association were noted in terms of frequencies or percentages. Continuous numerical data, for instance, age, gestational age at birth, and birth-weight were expressed as mean ± standard deviation. To check the association of Vertebrospinal Anomalies with study variables chi-square test was used.

REGISTRATION:

The study is registered with Clinical Trials and registration number is NCT06234020. Registration Date: 22/01/2024.

RESULTS:

We enrolled 106 patients in current study with mean age was 4.48±2.84 years and birth weight was found 2.58±0.52 kg as shown in Table 1. Out of 106 enrolled patients 62(58.5%) were male and 44(41.5%) patients were female. The male to female ratio was 1.41:1 in current study.

Table-1: Results of descriptive statistics of quantitative variables

Variables	N	Min	Max	Mean	SD
Age (years)	106	.50	13.00	4.48	2.84
Birth weight (kg)	106	1.50	5.00	2.58	.52
GA at birth (weeks)	106	30.00	40.00	36.96	2.28

We also found that just 62(58.5%) patients had normal birth weight and 74(69.8%) had SVD mode of delivery. In VACTERL association findings 36(34.0%) patients had vacterl association. The details of Krickenbeck Classification were given in Table-2.

The frequency of vertebro-spinal anomalies was found in current study as: 8(7.5%) patients had sacral agenesis, 4(3.8%) patients had scoliosis, 6(5.7%) patients had tethered cord and 2(1.9%) patients had thickened filum terminale. Overall frequency was 18.9%.

Table-2: Results of frequency distribution of qualitative variables

Variables		Frequency	Percent
sex	Male	62	58.5
	Female	44	41.5
Birth Weight Category	Normal Birth Weight	62	58.5
	Low Birth Weight	44	41.5
Mode of delivery	SVD	74	69.8
	LCSC	32	30.2
Vacterl Association	Yes	36	34.0
	No	70	66.0
Krickenbeck Classification	Anteriorly placed anus	2	1.9
	ARM with pouch colon	2	1.9
	No fistula	28	26.4

	Perineal fistula	18	17.0
	Rectobladderneck fistula	4	3.8
	Rectourethral fistula	14	13.2
	Rectovesical fistula	8	7.5
	Rectovestibular fistula	26	24.5
	Cloaca	4	3.8
Spinal Cord MRI findings	Normal cord	86	81.1
	Sacral agenesis	8	7.5
	Scoliosis	4	3.8
	Tethered cord	6	5.7
	Thickened filum terminale	2	1.9

The association of vertebro-spinal anomalies was associated significantly ($p<0.05$) with birth weight and VACTERL association. The detailed results were given in Table-3.

Table-3: Results of association of Vertebrospinal Anomalies with study variables

Variables		Spinal Cord MRI findings					Total	p value
		Normal cord	Sacral agenesis	Scoliosis	Tethered cord	Thickened filum terminale		
Sex	Male	46	6	2	6	2	62	0.108
		43.4%	5.7%	1.9%	5.7%	1.9%	58.5%	
	Female	40	2	2	0	0	44	
		37.7%	1.9%	1.9%	0.0%	0.0%	41.5%	
Birth Weight Category	Normal Birth Weight	52	2	4	2	2	62	0.047
		49.1%	1.9%	3.8%	1.9%	1.9%	58.5%	
	Low Birth Weight	34	6	0	4	0	44	
		32.1%	5.7%	0.0%	3.8%	0.0%	41.5%	
Mode of delivery	SVD	58	6	4	4	2	74	0.566
		54.7%	5.7%	3.8%	3.8%	1.9%	69.8%	
	LCSC	28	2	0	2	0	32	
		26.4%	1.9%	0.0%	1.9%	0.0%	30.2%	
Vacterl Association	Yes	18	8	2	6	2	36	0.0001
		17.0%	7.5%	1.9%	5.7%	1.9%	34.0%	
	No	68	0	2	0	0	70	
		64.2%	0.0%	1.9%	0.0%	0.0%	66.0%	

***Significant (p<0.05)**

DISCUSSION:

Out of our sample of 106 patients with ARM, almost one fifth of them had Vertebro-spinal Anomalies. This included both patients with basic ARM types and those with more complex ARM types. The screening regimen using ultrasound or MRI was adhered to in most individuals. The overall frequency was 18.9% of Vertebrospinal Anomalies.

The frequency of Vertebrospinal Anomalies in ARM patients in previous research ranged from 26% to 60%, which exceeds the frequency observed in our study.[16,18] Unlike our analysis, several other investigations only utilized MRI as a screening tool, potentially resulting in a higher incidence in those studies compared to ours. However, the occurrence rate in studies that do not follow a standardized screening approach may be low because of cases that were not correctly diagnosed and data that was not recorded. Additionally, there is no documentation available to determine if any additional imaging investigations were conducted.[16]

Oh et al[18] also conducted a study in which they found 22.6% spinal/vertebral systems anomalies in patients with Anorectal Malformation. While we found the 18.9% frequency of spinal/vertebral systems anomalies. In patients with ARM, these data will be useful for predicting spinal/vertebral systems anomalies. Every patient with ARM should undergo a comprehensive, systematic assessment.

Beaufort et al[14] evaluated the frequency of spinal cord abnormalities (SCA), particularly tethered spinal cord (TSC), in patients with ARM using spinal cord imaging techniques such as spinal cord ultrasound and MRI. SCA was detected in 22% of individuals with both simple and complex kinds of ARM. There were 8.1% patients with SCA who had TSC, and out of those, 4 patients had a neurosurgical operation without any complications. Consequently, it appears crucial to test for SCA in all patients with ARM, irrespective of the form of ARM. While we found 18.9% frequency of spinal/vertebral systems anomalies in which 5.7% had TSC. These results matched with current study results. The study found that the existence of VACTERL-association was independently linked to Vertebro-spinal Anomalies in multivariable analysis. Consistent with our findings, a prior study demonstrated a greater occurrence of Vertebrospinal anomalies in patients with ARM who also had VACTERL-association.[19] In addition, another study found a notable association between the degree of ARM and the existence of vertebro-spinal anomalies.[16] Our study and previous research have established associations, we have identified that SCA can occur in kids with any type of ARM. Therefore, it is recommended to test all children with ARM for vertebro-spinal anomalies. Our study has few limitations: duration of the study was short and study was based on single center study with limited sample size. Therefore we proposed that, in order to validate and expand our findings, a comparable comprehensive research may be conducted in multicentric settings with a bigger sample size to know more about the benefits of screening with ARM for vertebro-spinal anomalies.

CONCLUSION:

This study demonstrates that vertebrospinal anomalies are present in a significant proportion (18.9%) of patients with ARM. Among these, sacral agenesis was observed in 7.5% and tethered cord in 5.7% of cases. A statistically significant association was found between the presence of VACTERL association and vertebrospinal anomalies. These findings highlight the critical importance of incorporating routine spinal imaging into the diagnostic workup of all patients with ARM, regardless of the malformation subtype. Early identification of

such anomalies can facilitate timely neurosurgical consultation and optimize long-term functional outcomes

DECLARATIONS:

Ethics approval and consent to participate:

Ethical review board approval was taken from "Ethics research review board" of Pakistan Institute of Medical Sciences.

Approval No: F.3-1/2023(ERRB)/Chairman

All methods were performed in accordance with the ethical standards as laid down in the Declaration of Helsinki and its later amendments or comparable ethical standards.

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