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**ORIGINAL ARTICLE**

# Frequency of Vertebral Defects in patients with Gastrointestinal Anomalies

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## ABSTRACT

**Objective*:*** To show the frequency of vertebral defects associated with gastrointestinal anomalies.

**Study Design*:*** Descriptive Case study.

**Place and Duration of Study*:*** The study was conducted at the Radiology department of Children Hospital in collaboration with Paediatric Surgery, Pakistan Institute of Medical Sciences, Islamabad from October 2017 to August 2018.

**Material and Methods*:*** One hundred and eighty one patients (109 males and 72 females) were included in the study to find the frequency of associated vertebral defects in patients of upper and lower gastrointestinal anomalies. Upper gastrointestinal anomalies were confirmed through Barium swallow while children with lower GI anomalies underwent Distal Loopogram. X-ray Spine was done to rule out congenital spinal deformity.

**Results*:*** Out of 181 cases, 64 (35%) cases were identified with vertebral defects among all GI anomaly cases. Among the vertebral defects, coronal cleft vertebrae was found to be the most common (66.7%) deformity followed by Hemi-vertebrae (16%) while L5 and S1 vertebrae were observed to be most common site (n=18, 45%) of spina bifida in patients of lower GI anomalies.

**Conclusion:** Patients having vertebral defects associated with gastrointestinal anomalies should undergo a thorough physical and radiological examination to avoid misdiagnosis and incurable complications.

**Key Words*:*** *Gastrointestinal anomalies; Vertebral defects; Barium swallow; Spina bifida*

## INTRODUCTION

Gastrointestinal anomalies are usually uncommon malformations and surgical intervention is required for the survival of infants leading to a long term prognosis.[1] The incidence rate of gastrointestinal anomalies is 1:3 in 1000 live births, manifesting with or without associated anomalies.[2,3] The mortality rate for the infants having gastrointestinal anomalies associated with other malformations, is higher as compared to the gastrointestinal anomalies itself.[4] Gastrointestinal anomalies are categorized into upper and lower gastrointestinal anomalies. Esophageal atresia (upper GI anomaly) is a congenital anomaly causing the esophagus to become a blind ended

tube with or without tracheal fistula.[5] Literature has shown that the percentage of associated distal tracheal fistula is 86% while, in 7% cases, there is no fistula formation. The incidence rate of esophageal atresia is 1:25000.[6] Anorectal malformations (lower GI anomaly) are group of anomalies occurring in unborn baby with an absent or ectopic anus.[7] Boys are more affected as compared to girls.[8] The incidence rate of anorectal malformation varies between regions around the world but the reported rate is 2.0-2.5 per 10,000 live births.[9] According to Wingspread Classification, the anorectal malformations are categorized as high, intermediate and low type based on the level of rectal pouch relative to levator ani muscle (table 1).[10] Vertebral defects are commonly observed in patients having congenital gastrointestinal anomalies which includes, anorectal malformations (ARMs),[11] esophageal and duodenal atresia,[12,13] urinary tract diseases[14] and VACTREL anomalies.[15,16] VACTERL anomalies include vertebral defects, anal atresia, cardiovascular anomalies, tracheoesophageal fistula with esophageal atresia, renal and limb defects.[8] These anomalies can cause serious health problems even leading to death.[17] The prognosis of the patient depends on the severity of associated anomalies rather than the anorectal malformations or other GI anomaly itself.[18]

**TABLE 1: Wingspread classification of ARM**

| | Female | Male |
|---|---|---|
| High | Anorectal agenesis (with/ without rectovaginal fistula)<br>Rectal atresia | Anorectal agenesis (with/without rectoprostatic fistula)<br>Rectal atresia |
| Intermediate | Rectovestibular fistula Rectovaginal fistula<br>Anal agenesis without fistula | Rectobulbar urethral fistula<br>Anal agenesis without fistula |
| Low | Anovestibular fistula Anocutaneous fistula<br>Anal stenosis | Anocutaneous fistula<br>Anal stenosis |

Various studies have been conducted on association of gastrointestinal anomalies particularly; anorectal malformation with anal atresia, cardiovascular anomalies, tracheoesophageal fistula with esophageal atresia, renal and limb but there is a lack of research on its association with vertebral defects. The aim of this study was not only to show the frequency of vertebral defects associated with gastrointestinal anomalies but also to relate the incidence of these anomalies based on gender.

## MATERIAL AND METHODS

The study included 181 subjects, presented with clinical features of upper gastrointestinal anomalies such as inability to take feed and recoiling of nasogastric tube (NG tube) raising the suspicion of esophageal atresia. While, for lower gastrointestinal anomalies, all children without anal opening were considered in the study. Barium swallow was done to confirm upper gastrointestinal anomalies while children with lower GI anomalies underwent Distal Loopogram. X-ray Spine revealed congenital spinal deformity, ruled out initially through physical examination. All the infants included in the study had esophageal atresia and anorectal malformations. The study was conducted at Radiology department of Children Hospital in collaboration with Medicine department, Pakistan Institute of Medical Sciences, Islamabad. The data was analyzed through SPSS version 22.0.

## RESULTS

Mean age for disease was 7.04 ± 7.32 years. A total of 181 cases were identified with gastrointestinal anomalies, majority cases (60%) were males, and only 40% were females. Out of total cases, 140 (77%) infants had upper gastrointestinal anomalies whereas the cases of lower gastrointestinal anomalies were 41 (23%). The results have shown that the commonest type of ARM was found to be Intermediate type (44%) followed by High ARM (39%) and Low ARM (17%) respectively (fig 1)**.** While, 64 (35%) cases were identified with vertebral defects among all GI anomaly cases (table 2)**.**

Out of total 140 patients of upper GI anomalies, 24 (17%) cases were observed to have vertebral defects whereas out of 41 patients of lower GI anomalies, vertebral defects were found in 40 (97.5%) infants (table 3, 4)**.**

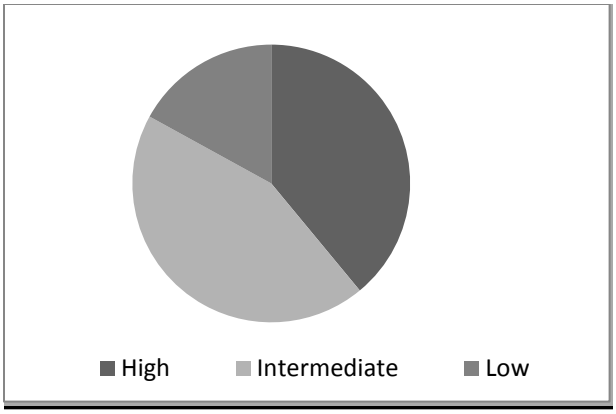


**Fig. 1: Percentage of types of Anorectal Malformation (ARM)**

**TABLE 2: Frequency of Vertebral defects in patients with gastrointestinal anomalies**

| | Frequency | Percentage | Cumulative Percentage |
|---|---|---|---|
| No | 117 | 64.6 | 64.6 |
| Yes | 64 | 35.4 | 100.0 |
| **Total** | **181** | **100.0** | |

**TABLE 3: Frequency of Vertebral Defects in Upper GI anomalies**

| | | Upper GI anomalies | |
|---|---|---|---|
| | | Yes | Total |
| Vertebral Defects | No | 116 | 116 |
| | Yes | 024 | 024 |
| **Total** | | **140** | **140** |

**TABLE 4: Frequency of vertebral defects in Lower GI anomalies**

| | | Lower GI anomalies | |
|---|---|---|---|
| | | Yes | Total |
| Vertebral Defects | No | 1 | 1 |
| | Yes | 40 | 40 |
| **Total** | | **41** | **41** |

The observed vertebral defects were coronal cleft, bifid, butterfly and hemi-vertebrae. Among these vertebral defects, coronal cleft vertebrae was found to be the most common (66.7%) deformity followed by hemi-vertebrae (16%) fig 2**.** L5 and S1 vertebrae were found to be the most common site (n=18, 45%) of spina bifida in patients of lower GI anomalies (table 5)**.**

**TABLE 5: Frequency of level of Spina Bifida**

| Level of Spina Bifida | Fre-quency | Percen-tage | Cumulative Percentage |
|---|---|---|---|
| L3 | 1 | 2.5 | 2.5 |
| L4 | 1 | 2.5 | 5.0 |
| L4, L5 | 2 | 5.0 | 10.0 |
| L5 | 6 | 15.0 | 25.0 |
| L5, S1 | 18 | 45.0 | 70.0 |
| L5, S1, S2 | 1 | 2.5 | 72.5 |
| S1, S2, S3 | 1 | 2.5 | 75.0 |
| S1 | 2 | 5.0 | 80.0 |
| S1, S2 | 5 | 11.0 | 92.5 |
| L4, L5, S1, S2 | 2 | 5.0 | 97.5 |
| S1, S5 | 1 | 2.5 | 100.0 |

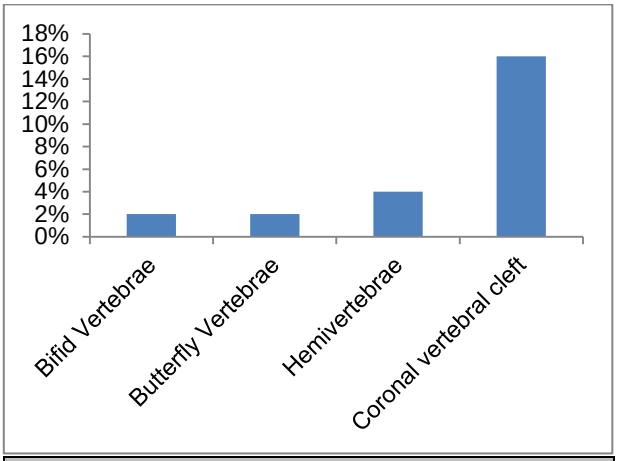


**Fig. 2: Percentage of types of Vertebral Defects**

Twenty eight (20%) patients were observed to have other deformities associated with upper GI anomalies which include anorectal malformations with and without recto-vesical fistula, with bucket handle deformity, ectopic location of anus, mal-rotation of gut, common cloaca and Meckel's diverticulum. Most common (35.7%) associated anomaly was found to be anorectal malformation without rectovesial fistula. Whereas, the observed cases having fistulous communication with lower GI anomalies were 6 (15%) consisting of rectovaginal, anorectal, recto-vesical and recto-urethral fistula. Recto-vesical and recto-urethral fistulas were found to be most common (33.3%) associated anomalies.

## DISCUSSION

Literature has shown that very few studies have been done on the occurrence of vertebral defects

in patients having upper and lower gastrointestinal anomalies. Esophageal atresia and anorectal malformations were observed to be the common findings in our patients of upper and lower GI anomalies respectively. Barium swallow and Distal Loopogram were done to confirm the diagnosis of upper and lower gastrointestinal anomalies, the suspicions of which were raised due to the feed intolerance and absence of anal opening. Out of total, 64 (35.4%) patients were observed to have vertebral defects that were confirmed with the aid of X-ray Spine. We conducted the study with the aim to show the frequency of vertebral defects in patients of gastrointestinal abnormalities. Association of different types of vertebral defects (24.1%) has also been observed in patients of esophageal atresia in a study done by SJ Keckler et al.[19]

FM Karrer also conducted a study showing the presence of vertebral defects in all patients having anorectal malformations.[20] There was only 28% incidence of vertebral defects in patients with anorectal malformations in a study done by SK Ratan et al. The observed defects were spina bifida and fusion anomalies of lumbosacral region.[17] Among them, the most common (66.7%) deformity was observed to be coronal cleft vertebrae followed by hemi-vertebrae (16%). This was in concord with our study where vertebral defects observed included spina bifida, coronal cleft, bifid, butterfly, hemi-vertebrae, lumbosacral agenesis and partial agenesis of sacrum.

Back in 1973, “Quan and Smith” named the other abnormalities and anomalies of Vertebral defects in particular, hemi-vertebrae and bifid vertebrae, Anal atresia, Tracheoesophageal fistula with Esophageal atresia, and Radial and renal dysplasia, as “VATER”.[21] In few studies, there was a strong evidence of occurrence of other musculoskeletal anomalies such as absent radius, additional or absent ribs, digital anomalies, hip dysplasia[19,22] while no such anomalies were observed in the current study.

In present study, the most common sites of spina bifida associated with anorectal malformation were L5 and S1 vertebrae. While sacrum was found to be the common site of vertebral defect in few studies[8,23,24] Mal-rotation of midgut, Meckel’s diverticula and pyloric stenosis are the anomalies associated with gastrointestinal anomalies in a review conducted by Holder et al.[25] Such findings were also obvious in our patients of esophageal atresia (except pyloric stenosis) for instance, anorectal malformations with and without recto-vesical fistula, with bucket handle deformity, ectopic location of anus, mal-rotation of gut, common cloaca and Meckel’s diverticulum. Of these associated anomalies, ARM without recto-vesical fistula was observed to be the common one. As a whole, 6 (15%) patients had fistulous communications associated with ARM leaving 85% with isolated ARM. Out of 6 patients, 2 had rectovaginal fistula, 2 had recto-urethral fistula while one had anorectal fistula and one patient was found to have recto-vesical fistula. Amongst them, recto-vesical and recto-urethral fistulas were the most common (33.3%) associated anomalies.

## CONCLUSION

Patients with upper and lower gastrointestinal anomalies, having esophageal atresia and anorectal malformations, were associated with multiple vertebral defects. In patients of esophageal atresia, coronal cleft vertebrae was found to be the most common finding followed by hemi-vertebrae while the most common sites of spina bifida associated with anorectal malformation were L5 and S1 vertebrae. Few patients of esophageal atresia were found to have other associated gastrointestinal anomalies as well, out of which, the common finding was ARM without recto-vesical fistula whereas recto-vesical and recto-urethral fistulas were observed to be the most common associated anomalies in patients with anorectal malformations. Most of the anomalies are life threatening and the patient’s recovery is mostly dependent on the severity of these associated anomalies rather than the gastrointestinal anomaly itself. Therefore, to avoid misdiagnosis and incurable complications, a thorough physical and radiological examination should be done to detect the disease in its early stages.

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