

## Vertebral anomalies associated with anorectal malformations incidentally found on contrast studies: a case series



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### ABSTRACT

**Background:** Spinal congenital malformations are closely associated with other congenital anomalies, are usually asymptomatic and be found incidentally. Moreover, spine abnormalities such as hemivertebra may cause further complications if left untreated. Among patients with VACTERL anomalies, spine anomalies account for 60-95% of them. This report aimed to describe rare and interesting cases of spinal malformation in children to provide further information on the importance of a proper approach in such cases.

**Case Presentation:** We present three pediatric cases of vertebral anomalies associated with anorectal malformations incidentally found in contrast studies. All patients have a history of anorectal abnormalities at birth and are referred to the hospital for contrast studies. The first case found semi-segmented hemivertebra at T8-9 levels, cleft at T10 level and high-type anorectal malformations (ARMs). The second case showed unsegmented hemivertebra at the level of L1 and butterfly vertebra at T10, approximately at the same level as the skin lesion, with the possibility of occult spina bifida and high-type ARMs. The third case revealed bilateral vesicoureteral reflux (VUR), grade 1 right hydronephrosis and left hydronephrosis, with semi-segmented hemivertebra at the level of L3, unsegmented hemivertebra at L4-5 and an extra lumbar vertebra, possibly a lumbosacral transitional vertebra (LSTV).

**Conclusion:** Thorough screening and other examinations such as contrast studies could reduce their morbidity as more sophisticated imaging may be done to better visualize the anatomical malformation.

**Keywords:** congenital anomalies, contrast studies, hemivertebra, spinal malformation.

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### INTRODUCTION

The vertebral column or the spine forms the central axis of the body's skeleton, which articulates with the skull cranially and the hip bones caudally. The vertebral column consists of seven cervical vertebrae, twelve thoracic vertebrae, five lumbar vertebrae, the sacrum and coccyx. Most spinal problems are caused by trauma, neoplasms and developmental anomalies.<sup>1</sup> Spinal congenital malformations are closely associated with other anomalies within and outside the spine, which can be identified as a coincidence association, namely VACTERL (vertebral defects, tracheoesophageal fistula, renal anomalies and limb abnormalities). The VACTERL syndrome is defined as the presence of three or more previous VACTERL anomalies, wherein the presence of spinal anomalies, the concurrence of other spinal abnormalities is as high as 15%-60%. Within those categories, the most common

concurrent anomalies are cardiac, urinary and gastrointestinal anomalies.<sup>2</sup>

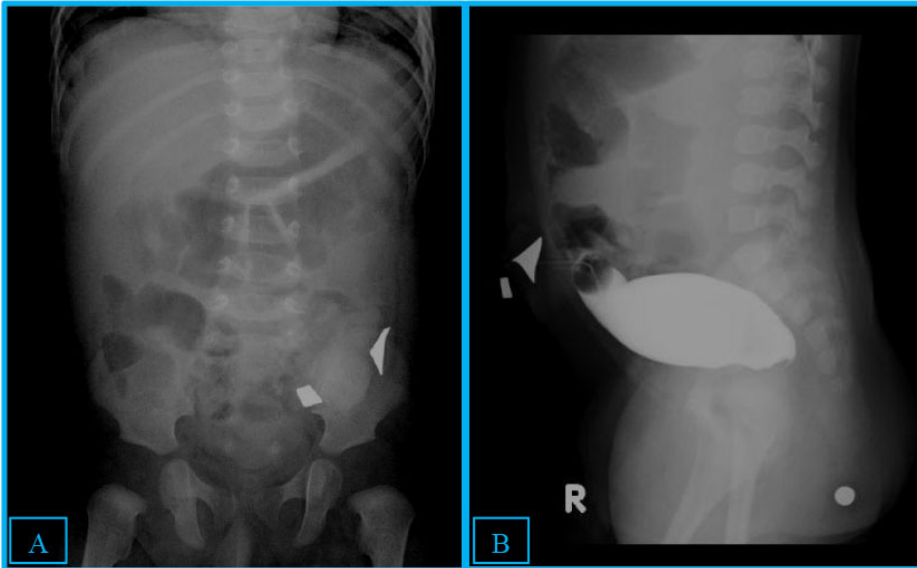
The anomalies in the VACTERL association develop at the different stages of embryogenesis. The vertebrae, especially the thoracic vertebrae, are the most often affected and formed as early as 23-32 days, while the heart, tracheoesophageal and forearm bones are the next at 29-41 days and anorectal structures being the latest at 45-56 days and beyond.<sup>3</sup> Bony parts of the vertebrae begin to form at around eight weeks of intrauterine life and the neuraxis completes its closure to form the neural tube. Abnormalities of the bony elements may present as the failure of formation, failure of segmentation or both in the form of mixed anomalies. These abnormalities cause an imbalance in the longitudinal growth of the spine, resulting in congenital spinal anomalies. As time goes by, patients with this deformity may have a higher risk of neurological complications.<sup>4</sup>

Despite of being closely associated with anorectal malformations other than urogenital anomalies, spine abnormalities may usually be asymptomatic and be found incidentally. In the case of detected vertebral or sacral anomalies through conventional radiograph, spinal Magnetic Resonance (MR) should be considered to visualize the more detailed anatomical structure, including the disc and endplates, which has prognostic and surgical implications.<sup>5-7</sup> We will describe a case series of vertebral anomalies incidentally found during contrast studies in pediatric patients with anorectal malformations to provide further information on the importance of a proper approach in such cases.

### CASE PRESENTATION

#### Case 1

A healthy four-month-old boy was brought to the department of radiology at Prof. Dr.



**Figure 1.** A) Plain abdominal radiograph AP projection showed incidental finding of asymptomatic semi-segmented hemivertebra at T8-9 levels and cleft at T10 level. B) Distal loopogram contrast study at lateral projection found the distance between the distal rectum to the anal dimple (round opaque marker) was approximately 4.7 cm with no evidence of additional contrast-filling tracts to surrounding organs, therefore concluded the diagnosis of high-type ARMs without the presence of a fistula.



**Figure 2.** A black-colored macular lesion with hair growing on its surface, raised the suspicion of a possible variant of faun-tail nevus in spina bifida.

I G. N. G. Ngoerah General Hospital-Bali for distal loopography contrast study. He was diagnosed with anal atresia at birth and surgery for colostomy formation was conducted soon afterward. He was referred by the department of pediatric surgery for posterior sagittal anorectoplasty (PSARP) planning. Other than this condition, he was otherwise healthy.

On the examination day, he was active, had normal vital signs, and had a non-

distended abdomen. On the stoma site, traces of fecal material looked normal and no sign of infection was present.

On plain AP projection abdominal radiograph prior to the contrast study, semi-segmented hemivertebra was discovered at T8-9 levels and cleft at T10 level (Figure 1A). The distal loopogram study showed normal descending colon and rectosigmoid mucosal linings, no evidence of additional tracts to

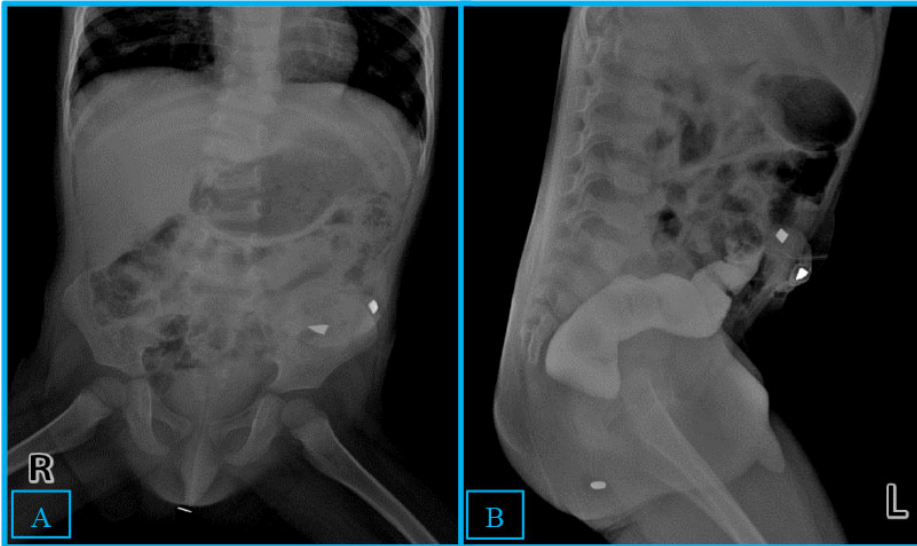
surrounding organs, and the distance measured between the distal rectum to anal dimple concluded high-type anorectal malformations (ARMs) without the presence of fistula (Figure 1B). After the contrast examination, the patient went home with no sign of allergic reaction due to contrast agents.

### Case 2

A two-year-old boy was referred from a hospital in the rural area for distal loopography study at Prof. Dr. I G. N. G. Ngoerah General Hospital-Bali. He was first discharged as a “well baby” after being observed by a primary care health service provider soon after birth. On the day after, he was bloated and vomiting profusely, and his parents brought him to the hospital. After a series of examinations, the doctor diagnosed him having anal atresia and conducted urgent surgery for colostomy the day after. However, the lack of facilities and difficulties accessing the state hospital in rural areas due to pandemics delayed further PSARP procedure planning.

On the day of examination, it was observed that he was active, had normal vital signs, no gait disturbances, and had a common developmental milestone record. However, we discovered a black-colored macular lesion on his back measured approximately 3 x 1.5 cm with hair growing on its surface (Figure 2). His parents said that it was a birthmark present since his birth with no evidence of abnormal growth of the skin lesion. On the stoma site, traces of healthy-looking fecal materials were seen and no evidence of infection was present.

On plain abdominal radiograph prior to the contrast study, we revealed evidence of unsegmented hemivertebra at the level of T12-L1 and butterfly vertebra at T10 approximately at the same level of the skin lesion (Figure 3A), with the possibility of occult spina bifida. The distal loopogram study showed normal descending colon and rectosigmoid mucosal linings, no evidence of additional tract to surrounding organs, and the distance measured between the distal rectum to anal dimple concluded high-type anorectal malformations (ARMs) without the presence of fistula (Figure 3B). After the contrast examination, the patient



**Figure 3.** A) Plain abdominal radiograph AP projection showed incidental findings of asymptomatic mixed-type vertebral anomalies consisting of unsegmented hemivertebra at the level of T12-L1 and butterfly vertebra at T10. B) Distal loopogram contrast study at left lateral projection found the distance between distal rectum to the anal dimple (round opaque marker) was approximately 3.5 cm with no evidence of additional tracts to surrounding organs, therefore concluded high-type ARMs with no fistula.



**Figure 5.** Abdominal AP radiograph the day after the examination showed a semi-segmented hemivertebra at the level of L3, an unsegmented hemivertebra at L4-5 and an extra lumbar vertebra, possibly a lumbosacral transitional vertebra (LSTV) resulting lumbar dextroscoliosis.



**Figure 4.** A) AP projection of pelvic radiograph revealed slightly-oblique oriented pelvic bone. B,C) Contrast study resulted in bilateral VUR and grade 3 right hydroureteronephrosis with no evidence of contrast-filling fistula.

was discharged with no complications regarding the contrast study.

**Case 3**

A ten-year-old girl was referred to the department of radiology at Prof. Dr. I G. N. G. Ngoerah General Hospital-Bali by a pediatric surgeon for a retrograde urocytography examination, with the suspicion of her having urogenital tract fistula. She was first diagnosed with anorectal malformations at birth and had surgery for colostomy the day after. On her second birthday, she had posterior sagittal anorectal vaginal urethral plasty (PSARVUP) surgery and since then has had normal defecation through the anal

canal made surgically. However, her micturition was never normal, as she complained it was constantly leaking, and she never had the urge to urinate. Other than the malformations described before, she was informed that she had congenital talipes equinovarus (CTEV) at birth, treated with repositioning and splinting,

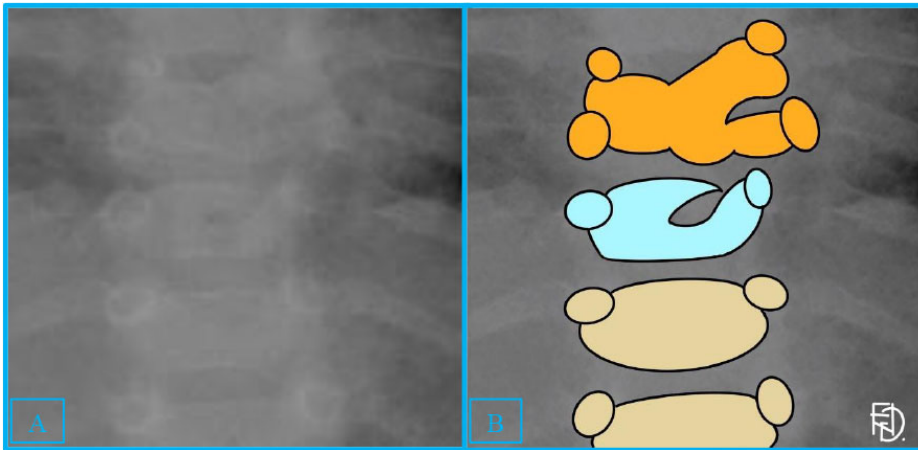
On the day of examination, we revealed an active girl with normal vital signs, walking slowly in a slightly slanted body. The physical examination for the other organs was unremarkable.

On plain AP projection pelvic radiograph prior to the contrast study, we noticed a slightly-oblique oriented pelvis, which raised our suspicion of scoliosis

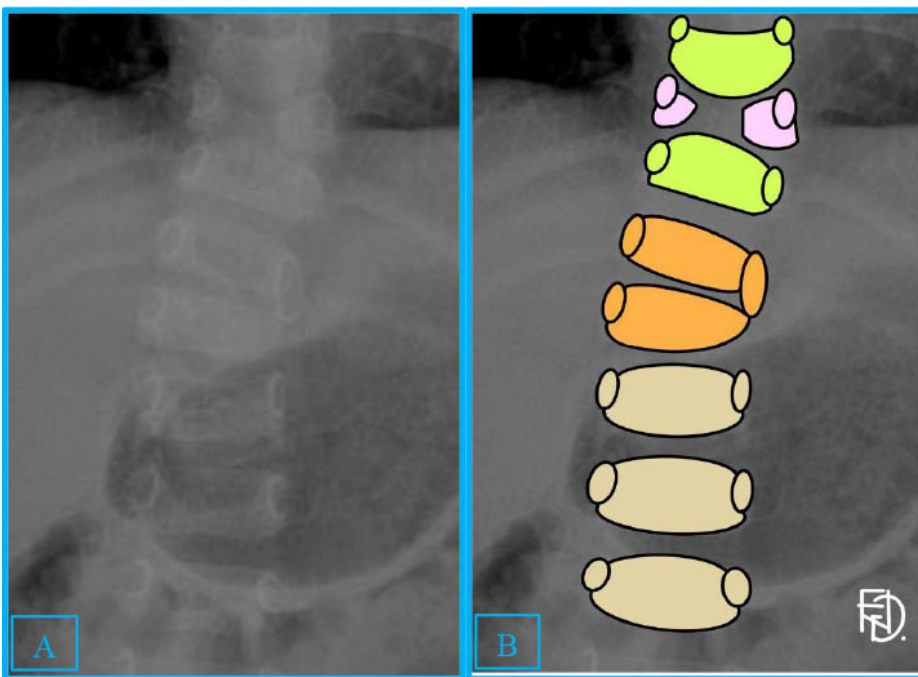
(Figure 4A). The urocytography resulted in bilateral vesicoureteral reflux (VUR), grade 1 right hydroureteronephrosis and left hydroureter, and no evidence of contrast-filling fistula in the current examination (Figure 4B and C). To have a better visualization of the vertebra, we conducted another imaging study on the day after the examination and revealed a semi-segmented hemivertebra at the level of L3, an unsegmented hemivertebra at L4-5 and an extra lumbar vertebra, possibly a lumbosacral transitional vertebra (LSTV) (Figure 5). The patient's parents said that this anomaly was never mentioned before, and now she is still having regular outpatient visits for her conditions.

**DISCUSSION**

Embryologically, spine formation is divided into four overlapping stages: mesenchymal, chondrification, primary and secondary ossification. The development of the spine starts at the time with the expansion of the notochord. The



**Figure 6.** Enlarged view (A) and schematic diagram of the first case (B) showed semi-segmented hemivertebra at T8-9 levels in orange and cleft at T10 level in light blue.



**Figure 7.** Enlarged view (left) and schematic diagram of the second case (right), showing butterfly vertebra (pink) at T10, with increased endplate convexities of the upper and lower vertebrae (light green), also unsegmented hemivertebra at the level of T12-L1 (orange).

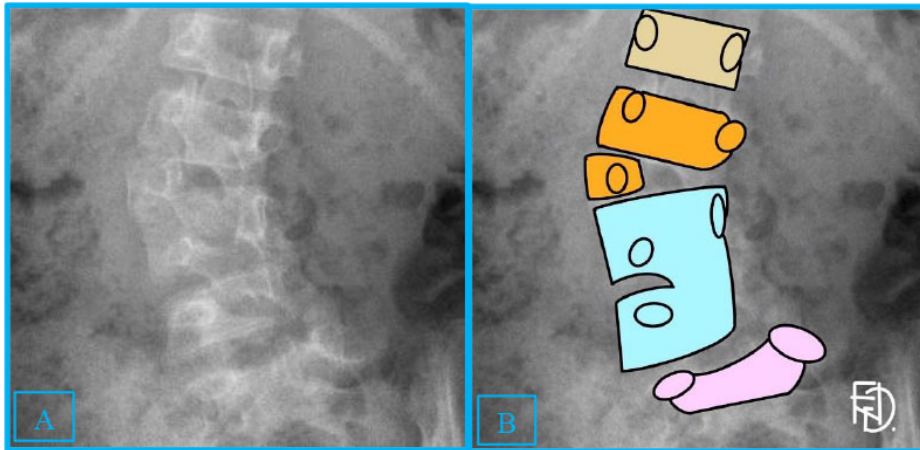
mesoderm surrounding the notochord is divided into three areas: the paraxial, lateral and intermediate. The intermediate mesoderm contributes to the urogenital system; the lateral mesoderm contributes to the cardiac, pulmonary and other organ systems. The paraxial mesoderm develops into 42 to 44 pairs of somites in a craniocaudal direction. Sacral elements are the last to appear at about day 31 of intra-uterine life. Each somite then differentiates into a dermomyotome and a

sclerotome.<sup>8</sup> The sclerotome then becomes the bony part of the vertebral column. The 31st sclerotome represents the L1-L2 level, considering that every vertebra forms from the fusion of the superior sclerotome's caudal half with the inferior sclerotome's cranial half. At week three to six, two lateral chondrification centers appear, forming the cartilaginous vertebral column. Simultaneously, the notochord degenerates at the level of vertebral bodies and remains only at the intervertebral level

to create the nucleus pulposus. Ossification of the vertebra occurs in three primary centers and five secondary centers. One primary center is in the centrum, and the remaining two are on each side of the neural processes. Secondary ossification centers are, respectively tip of the spinous process, the tip of both transverse processes and superior and inferior surfaces of the vertebral bodies.<sup>6,8</sup>

Compared with other abnormalities stated in VACTERL, the most common defect was reported in vertebral anomalies in 60-95% of VACTERL patients.<sup>8</sup> According to Kaplan et al., malformations of the spine can further be classified into three groups: neural tube defects (spina bifida), defects of segmentation (block vertebra) and defects of formation (hemivertebra and butterfly vertebra).<sup>9</sup> All of our three cases (Figure 6, 7, 8) reflect the defect in the formation and only case 3 (Figure 8) shows a defect in segmentation as stated in the study by Mohanty et al., failure of formation was the most commonly found vertebral anomaly.<sup>4</sup>

Hemivertebra is a congenital spine deformation where only half of the vertebral bodies develop. The incidence of hemivertebra is estimated at 0.1-1.0 per 1,000 birth and is reported to be more common in females, with a male-to-female ratio of 0.31. It can be fully segmented, semi-segmented or non-segmented. Segmented hemivertebra still have growth plates cranially and caudally. If fusion occurs with the cranial and caudal vertebral bodies, they create semi-segmented vertebra. A non-segmented vertebra has not separated from either the cranial or caudal vertebra.<sup>9,10</sup> Hemivertebra is also the most common cause of congenital scoliosis since a segmented hemivertebra can lead to spinal deformity due to unbalanced spinal growth if left untreated.<sup>11</sup> While our three cases show incidental findings of hemivertebra, our third case signifies a more complex case causing lumbar dextroscoliosis by semi-segmented hemivertebra at the level of L3, unsegmented hemivertebra at L4-5 and a possible LSTV. Unfortunately, while having difficulties in controlling the progression of the deformity in a growing spine, the timing of the surgery is also essential. As stated in a study by



**Figure 8.** Enlarged view (left) and schematic diagram of the third case (right) semi-segmented hemivertebra at the level of L3, unsegmented hemivertebra at L4-5 and an extra lumbar vertebra, possibly a lumbosacral transitional vertebra (LSTV) resulting lumbar dextroscoliosis.

Bagheri et al., the fusion of young children should be conducted at the age of eight to ten years old.<sup>12</sup> Figure 6 and 8 show a schematic and respective diagram of our first and third cases showing various types of hemivertebra.

As shown in our second case, the butterfly vertebra is a deformity characterized by the congenital failure of convergence of the chondrification centers of the vertebral bodies. The deformed vertebra consists of two partially or entirely separated hemivertebrae, divided by a persistent cartilaginous septum. The predominant region is the lumbar spine, followed by the thoracic region. As mentioned in a case report by Kapetanakis et al. and Hopkins et al., the defect is usually asymptomatic and incidentally detected.<sup>6,13</sup> Our second patient also showed a cutaneous lesion, which could signify an underlying spine and spinal cord anomaly. Faun tail nevus, which is also known as simple nevoid hypertrichosis, is famous for being a marker for spinal dysraphism, as described in a case report by Yamini et al.<sup>14</sup> Other than being asymptomatic, butterfly vertebra could also signify another more serious disease such as Alagille syndrome, in which they had the abnormal liver function in concurrence with other skeletal and facial anomalies.<sup>15</sup>

Imaging studies have a pivotal role in making a correct diagnosis. Therefore, the proper therapeutic means could be initiated. In the case of vertebral

anomalies, other imaging means for screening should be done to seek after VACTERL association.<sup>2</sup> Abdominal and pelvic ultrasonography (US), performed with an 8-MHz transducer, in which this technique facilitates optimal screening of the entire urinary tract including the bladder, bladder neck, and posterior urethra in boys. The spinal US can usually be done to detect spinal dysraphism. Spinal Magnetic Resonance Imaging (MRI) is the imaging modality of choice for better visualization of soft tissue-related spine disease and is actually recommended in patients with vertebral anomalies, sacral dysplasia, or abnormal results at the spinal US.<sup>5,16</sup> The timing of MRI depends on the clinical presentation of the patients with congenital spinal deformity. A child below the age of five years may warrant MRI under general anesthesia. Hence, in the neurologically intact child with no features of cord anomalies, MRI can be deferred until the child is able to tolerate the investigation.<sup>4</sup>

## CONCLUSION

Vertebral anomaly is one of the most commonly found congenital malformations and is frequently associated with anorectal malformations. Therefore, it is important to carefully screen vertebral malformations simultaneously with other examinations, especially contrast studies. As most cases are asymptomatic and incidentally found on X-ray, thorough

assessment and further workups could reduce their morbidity and prevent complications in the future.

## CONFLICT OF INTEREST

The authors declare no conflicts of interest.

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The authors received no external funding.

## AUTHOR CONTRIBUTION

All the authors equally contributed to the study, from the conceptual framework, data gathering, and analysis to interpreting the study results.

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