Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Medicine

Early-Onset Congenital Scoliosis Revealing Multilevel Congenital Vertebral Segmentation Anomalies Associated with Suspected Horseshoe Kidney: A Pediatric Case Report

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DOI: https://doi.org/10.36347/sjmcr.2026.v14i06.050 | **Received:** 04.05.2026 | **Accepted:** 13.06.2026 | **Published:** 22.06.2026

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Abstract **Case Report**

Introduction: Congenital vertebral segmentation anomalies are a heterogeneous group of developmental malformations that may lead to progressive spinal deformity during growth. They are frequently associated with extra-spinal abnormalities, particularly involving the genitourinary system; therefore, systematic screening for associated visceral, especially renal, anomalies is warranted. ***Case presentation:*** We report the case of a 7-year-old girl referred for evaluation of a progressive lumbar spinal deformity. Clinical examination revealed trunk asymmetry, pelvic obliquity, and a moderate lumbar paravertebral prominence. Standing full-spine radiographs demonstrated multilevel vertebral segmentation anomalies with vertebral blocks extending from L1 to L5, associated with mild congenital lumbar scoliosis. Spinal magnetic resonance imaging confirmed the vertebral abnormalities and excluded spinal cord and conus medullaris involvement. It also showed an empty left renal fossa, suggesting an associated renal developmental anomaly compatible with a suspected renal fusion anomaly. Abdominal ultrasonography confirmed the absence of a kidney within the left renal fossa. During two years of follow-up, moderate progression of the spinal deformity was observed without neurological impairment. ***Conclusion:*** This case highlights the importance of systematic screening for associated visceral anomalies in children with congenital vertebral malformations. It also emphasizes the need for multidisciplinary management and long-term clinical, functional, and radiological follow-up to monitor deformity progression throughout growth.

Keywords: Congenital vertebral malformations; vertebral block; congenital scoliosis; horseshoe kidney; pediatric rehabilitation.

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I. INTRODUCTION

Congenital vertebral malformations constitute a heterogeneous group of developmental anomalies resulting from disturbances in the formation, segmentation, or resegmentation of the vertebral precursors during embryogenesis. These abnormalities occur early, between the third and eighth weeks of gestation, and mainly include hemivertebrae, unsegmented bars, vertebral blocks, and mixed anomalies. Their clinical consequences vary according to the type, location, and extent of the anomaly, ranging from incidental findings to severe progressive spinal deformities [1].

Congenital vertebral anomalies represent one of the leading causes of congenital scoliosis. The natural history of these deformities depends largely on the growth potential of the abnormal vertebral segments and the resulting imbalance between the convex and concave sides of the spine. Multilevel segmentation defects, particularly vertebral blocks, may alter normal spinal growth and progressively lead to complex three-dimensional deformities affecting trunk balance and posture during childhood and adolescence [2].

Because the vertebral column and several visceral organs originate from closely related embryological structures, congenital vertebral malformations are frequently associated with anomalies involving other organ systems. Renal abnormalities are among the most commonly reported associated malformations. Cardiovascular, spinal cord, and genitourinary anomalies may also coexist, highlighting

the importance of a comprehensive diagnostic evaluation in affected children [3].

The coexistence of extensive vertebral segmentation defects and renal developmental anomalies remains uncommon and illustrates the complex embryological relationships underlying congenital malformations. Such associations reinforce the need for systematic screening for extra-spinal abnormalities and multidisciplinary long-term follow-up [4].

We report the case of a child presenting with early-onset congenital scoliosis secondary to extensive multilevel vertebral segmentation anomalies associated with a suspected renal fusion anomaly compatible with a horseshoe kidney.

II. CASE PRESENTATION

Medical history

A 7-year-old girl was referred to our Physical and Rehabilitation Medicine department for evaluation of a progressive lumbar spinal deformity identified on radiological examination. The deformity had initially been noticed by her mother during early childhood as a subtle lumbar asymmetry, which gradually became more apparent by the age of six years.

The obstetric history revealed a pregnancy complicated by polyhydramnios, carried to term, followed by uncomplicated vaginal delivery with a birth weight of 3.6 kg. Psychomotor development was within the normal range for age. No parental consanguinity, family history of spinal deformity, or congenital malformation was reported.

Initial clinical examination

At the initial evaluation, the patient had a slender habitus, measuring 118 cm in standing height and 58 cm in sitting height, with a body weight of 18 kg. Clinical examination revealed mild trunk asymmetry, characterized by left scapular prominence, a depressed left shoulder, flattening of the right waist crease, and left pelvic obliquity.

Frontal balance assessment revealed a 0.5 cm deviation of the C7 plumb line to the right. Sagittal alignment measurements showed an occiput-to-wall distance of 2 cm, a C7-to-wall distance of 3 cm, a D6-to-wall distance of 0 cm, an L3-to-wall distance of 0.5 cm, and an S2-to-wall distance of 0 cm.

The Adams forward bending test revealed a mild right lumbar prominence estimated at 3°. Spinal palpation elicited no pain but revealed a left lumbar paravertebral muscle contracture. Overall spinal mobility was preserved, with a finger-to-floor distance of 0 cm and a Schober index of +3 cm. A 4 mm right-sided lower limb length discrepancy was also documented.

Paraclinical assessment

Standing full-spine radiographs demonstrated multilevel vertebral segmentation anomalies characterized by stepped vertebral blocks involving L1-L2 and L3-L4-L5. These abnormalities were associated with mild lumbar scoliosis with left concavity and pelvic obliquity of 2.8 mm.

Spinal MRI ruled out spinal cord and conus medullaris abnormalities and confirmed the vertebral segmentation defects. In addition, MRI showed an empty left renal fossa, suggesting an associated renal developmental anomaly compatible with a suspected renal fusion anomaly. Abdominal ultrasonography confirmed the absence of a kidney within the left renal fossa, while the right kidney displayed normal morphology and size.

The patient was placed under regular clinical and radiological monitoring and was referred for pediatric assessment and further etiological investigations to evaluate the possibility of an associated polymalformative syndrome.

Two-year follow-up

The patient was managed conservatively with regular clinical and radiological follow-up. At the age of 9 years, follow-up assessment showed moderate progression of the deformity. Standing height reached 128.5 cm, sitting height was 62 cm, and body weight was 23 kg.

Clinical examination revealed progression of the lumbar prominence to 4° on the Adams test, a finger-to-floor distance of 5 cm, and a Schober index reduced to +2 cm. Sagittal assessment showed mild deterioration of spinal balance, including a D6-to-wall distance of -1 cm.

Follow-up standing radiographs confirmed progression of the segmentation anomaly, with a vertebral block extending from T12 to L4, including a continuous block from L1 to L4. No posterior vertebral wall abnormality was identified. Imaging also showed relative thoracic hypokyphosis and a thoracolumbar scoliotic posture with right convexity. Throughout follow-up, the neurological examination remained normal, with no evidence of motor, sensory, or sphincter dysfunction.

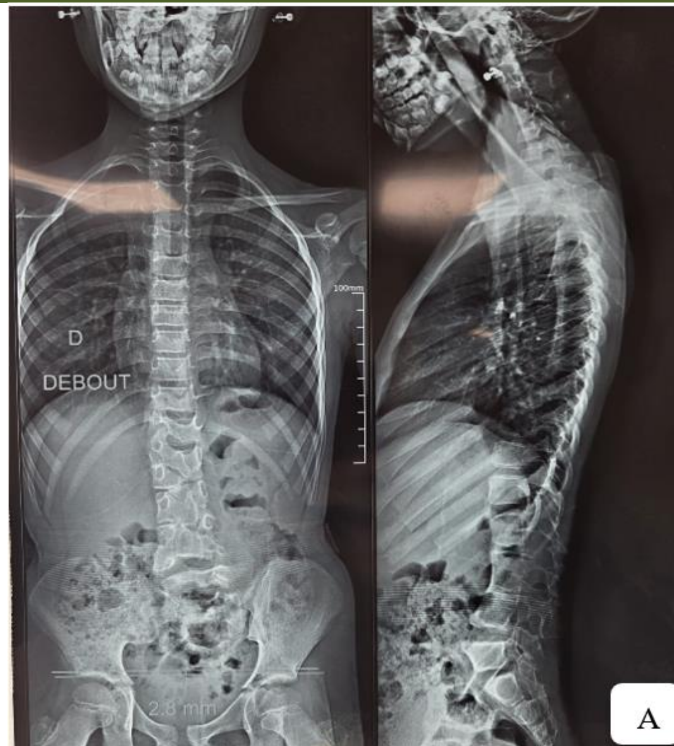

Figure 1: Standing full-spine radiographs obtained at age 7 years (A) and age 9 years (B)

Progression of multilevel vertebral segmentation anomalies with congenital thoracolumbar deformity during follow-up.

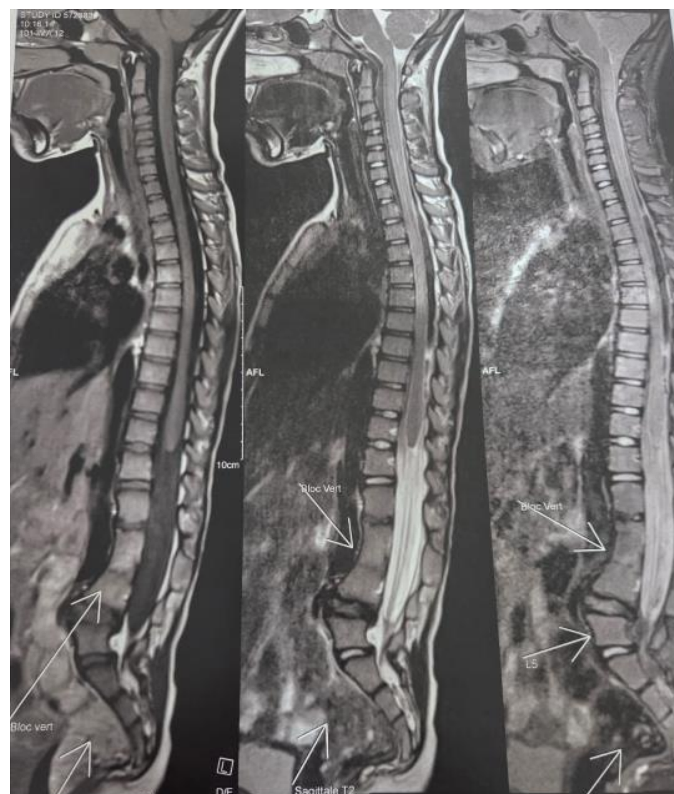

Figure 2: Spinal magnetic resonance imaging (MRI)

MRI confirming multilevel vertebral segmentation anomalies and demonstrating the absence of spinal cord or conus medullaris abnormalities.

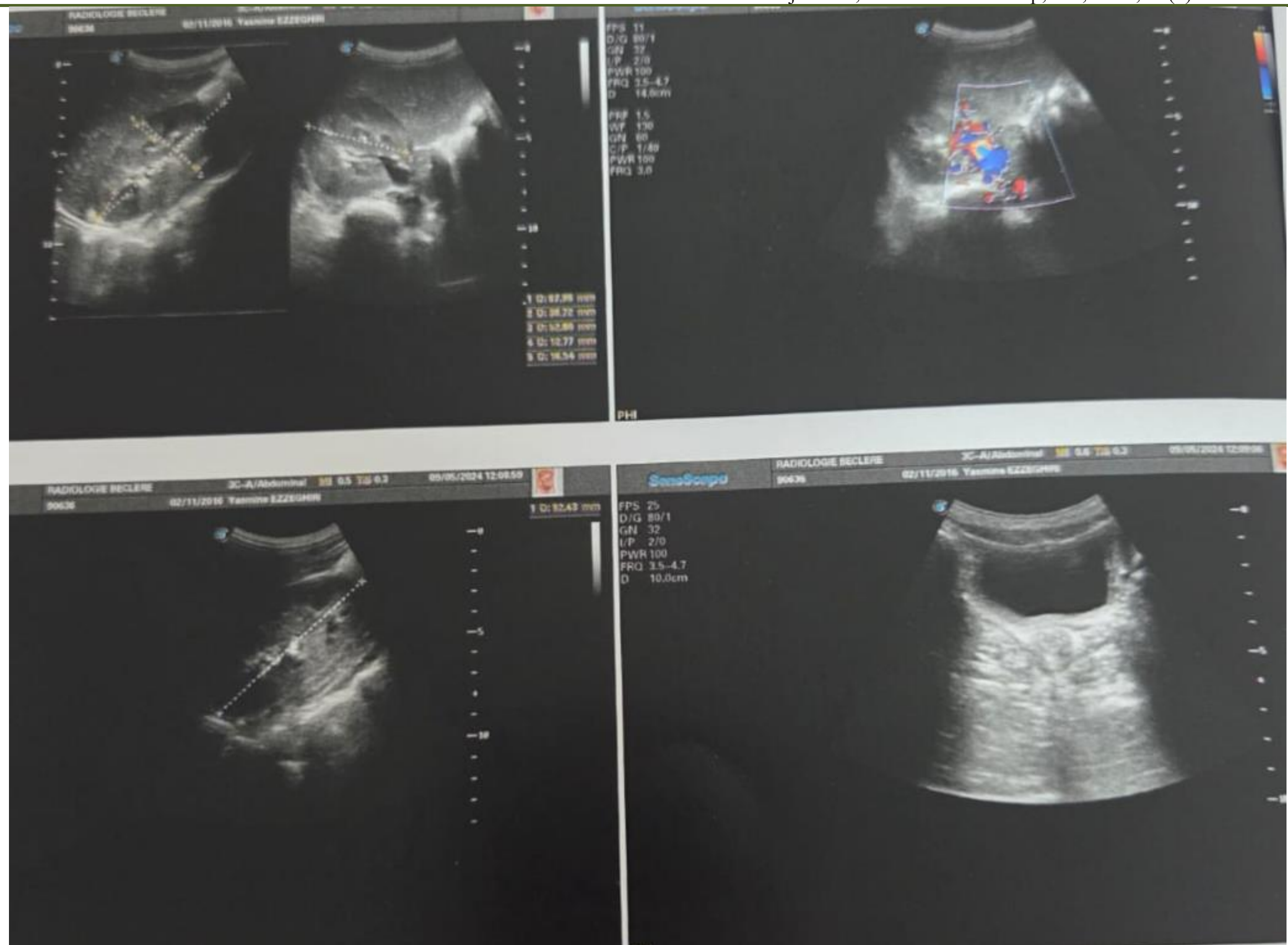

Figure 3: Abdominal ultrasonography

Ultrasound image showing the absence of a kidney within the left ren[illegible]a, suggestive of an associated renal developmental anomaly.

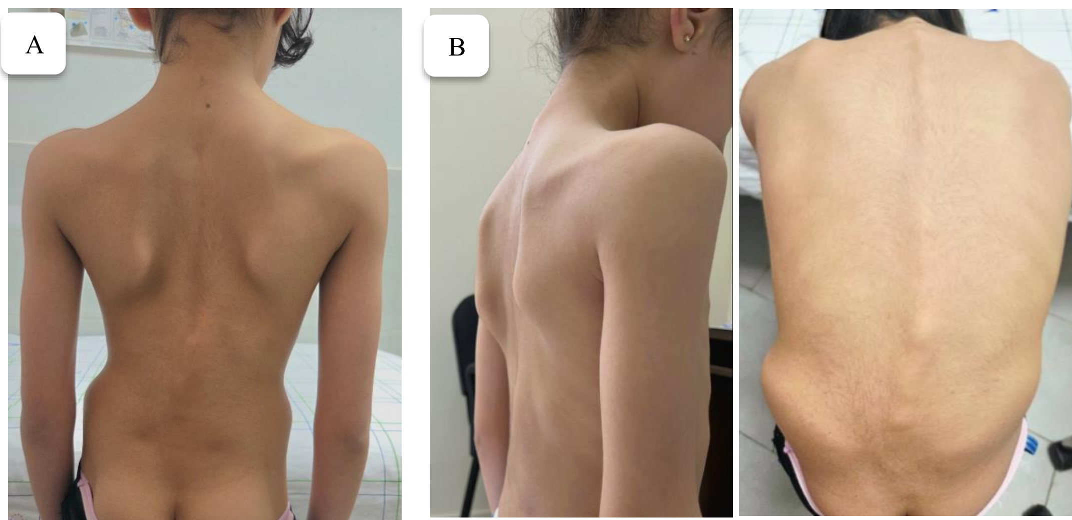

Figure 4: Clinical photographs of the patient
(A) Standing frontal view demonstrating trunk asymmetry and waistline imbalance
(B) Standing lateral view showing relative thoracic hypokyphosis
(C) Forward bending test (Adams test) demonstrating a thoracolumbar prominence

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III. DISCUSSION

Congenital vertebral segmentation anomalies result from disturbances in somitic resegmentation during early embryogenesis, leading to partial or complete fusion of adjacent vertebral bodies with secondary growth asymmetry [1]. Depending on their location and extent, these defects may alter spinal growth and induce progressive deformities throughout childhood. Multilevel vertebral blocks are relatively uncommon and are generally associated with a higher risk of spinal imbalance than isolated segmentation defects because of their cumulative effect on longitudinal growth [4].

Congenital scoliosis secondary to vertebral segmentation anomalies results from asymmetric longitudinal growth of the spinal column. Although fully segmented hemivertebrae are traditionally considered the most progressive congenital deformities, extensive multilevel vertebral blocks may also significantly disturb spinal alignment by restricting growth across multiple motion segments. The risk of progression depends on several factors, including the type and location of the anomaly, the symmetry of the block, the number of involved vertebrae, associated malformations, and the child's remaining growth potential [2,3].

In the present case, the involvement of multiple lumbar vertebrae extending initially from L1 to L5, with subsequent radiological progression to include T12, represents an extensive form of congenital segmentation anomaly. Although the deformity remained clinically moderate and the neurological examination was normal, the documented progression highlights the need for continued surveillance, particularly during periods of accelerated growth such as puberty, when curve progression may become more pronounced [2,5].

The association between vertebral and renal anomalies is well established and can be explained by their closely related embryological origins. During early embryogenesis, the vertebral column and urinary system develop from adjacent mesodermal structures. Disturbances occurring during this critical developmental period may therefore affect both systems, explaining the relatively high prevalence of genitourinary anomalies reported in patients with congenital vertebral malformations [3].

A recent meta-analysis reported an overall incidence of genitourinary anomalies of approximately 20-25% in patients with congenital scoliosis, including renal agenesis, ectopic kidney, duplication anomalies, and renal fusion defects [3]. Horseshoe kidney is the most frequent renal fusion malformation, with an estimated incidence of approximately 1 in 400 births [6]. In our patient, abdominal ultrasonography demonstrated the absence of a kidney within the left renal fossa, while MRI findings suggested an associated renal developmental anomaly compatible with a suspected horseshoe kidney. Additional imaging would be required for definitive anatomical characterization, but this finding reinforces the importance of systematic screening for associated renal abnormalities in children with congenital vertebral malformations [7].

From a genetic perspective, increasing evidence highlights the molecular complexity underlying congenital vertebral anomalies. Mutations involving genes associated with somitogenesis and vertebral segmentation, including NOTCH pathway-related genes, DLL3, and MESP2, have been implicated in a variety of congenital spinal malformation syndromes [8]. Although genetic testing was not performed in our patient, it should be considered in cases presenting with multiple congenital anomalies or suspected syndromic associations.

Imaging plays a central role in both diagnosis and follow-up. Standard radiographs remain the cornerstone for identifying segmentation defects and monitoring spinal deformity progression. MRI is essential for detecting associated spinal cord abnormalities, which may coexist in congenital scoliosis [9]. Abdominopelvic ultrasonography remains a first-line screening examination for renal malformations in this context [3]. Recent advances in three-dimensional imaging, morphometric analysis, and predictive models may further improve risk stratification and help individualize follow-up strategies during growth [10].

Management of congenital vertebral segmentation anomalies depends on the severity of the deformity, its progression rate, and the child's remaining growth potential. Stable deformities may be managed conservatively with regular clinical and radiological surveillance, whereas progressive forms may require orthopedic or surgical treatment, particularly in cases of rapid progression or major trunk imbalance. Available surgical options include hemivertebra resection, short-segment fusion, growth-friendly instrumentation, and guided-growth techniques aimed at controlling deformity progression while preserving spinal growth [2,10].

From a Physical and Rehabilitation Medicine perspective, management extends beyond radiological monitoring. Functional assessment is essential to evaluate posture, trunk balance, spinal mobility, gait, and potential limitations in daily activities. Rehabilitation physicians play a key role in longitudinal follow-up, early identification of functional deterioration, patient and family education, and coordination of multidisciplinary care.

In our case, the absence of neurological deficit, the relatively moderate magnitude of the deformity, and the preservation of functional abilities supported a conservative approach based on close clinical and radiological surveillance. Nevertheless, the documented

progression underscores the inherent growth-related risk associated with extensive multilevel segmentation anomalies and justifies continued monitoring, with reassessment of treatment options if progression accelerates during puberty.

This clinical case highlights the importance of a multidisciplinary approach involving pediatricians, rehabilitation physicians, radiologists, nephrologists, and spine surgeons. Such collaboration allows early diagnosis of associated malformations, appropriate surveillance, and optimization of long-term functional outcomes.

Conclusion

Congenital multilevel vertebral segmentation anomalies are uncommon developmental disorders that may lead to progressive spinal deformity throughout growth. This case illustrates the diagnostic and prognostic challenges posed by extensive vertebral block anomalies associated with a suspected renal fusion anomaly.

The coexistence of vertebral and renal malformations highlights the importance of systematic screening for associated anomalies in all children diagnosed with congenital scoliosis. Early identification of extra-spinal abnormalities is essential to ensure appropriate multidisciplinary management and long-term surveillance.

Although the deformity remained moderate and neurologically asymptomatic during follow-up, the documented progression confirms the growth-related evolutionary potential of multisegmental vertebral anomalies. Careful clinical, functional, and radiological monitoring throughout childhood and adolescence is therefore required to detect progression early and guide timely therapeutic decision-making.

This observation emphasizes the value of a multidisciplinary approach involving pediatricians, rehabilitation physicians, radiologists, nephrologists, and spine surgeons in order to optimize functional outcomes and anticipate potential complications during growth.

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