CASE REPORT

Spondylocostal Dysostosis: Mild variant in a Nigerian Newborn

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ABSTRACT

Background: Spondylocostal dysostosis (Jarcho-Levin Syndrome) is a rare congenital abnormality of spines and ribs which usually presents with trunk dwarfism, scoliosis, and respiratory symptoms. Its occurrence can be sporadic or familial. The clinical characteristics are vertebral abnormalities of hemivertebrae, butterfly vertebrae, scoliosis and variable rib abnormalities. Imaging is critical in its evaluation and management. **Case Presentation:** A 4-day-old child of a non-consanguineous couple delivered with swelling at the back. The family history was unremarkable for congenital anomalies. Clinical examination and radiological findings was consistent with spondylocostal dysostosis. **Conclusion:** Spondylocostal dysostosis is a rare musculoskeletal abnormality. Imaging plays a vital role in its diagnosis. An early diagnosis is necessary to prognosticate and institute appropriate management.

Key words: Spondylocostal dysostosis, Jarcho-Levin, Hemivertebrae, Rib anomalies, Congenital scoliosis.

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Introduction

Spondylocostal dysostosis (SCD) also known as spondylocostal dysplasia or Jarco-Levin syndrome is a rare congenital disorder associated with vertebral and rib abnormalities.[1,2] It presents as one of the spectra of short trunk dysplasias with hereditary or sporadic occurrence. It is usually inherited as an autosomal recessive or rarely autosomal dominant trait.[3] The clinical features include short neck and trunk, scoliosis, dyspnea with sometimes respiratory failure due to constricted thorax.[1,2]

Hemivertebra is a congenital vertebral anomaly that results from failure of development of one-half of the vertebral body and is reported to be the common cause of congenital scoliosis with an estimated incidence of 0.5-1.0 per 1,000 births; it was also reported to be more common in females fetuses, with a male to female ratio of 0.68 for solitary vertebral anomalies and 0.31 for multiple vertebral anomalies.[4] Congenital malformations of the vertebral bodies are recognised to be associated with other diseases/syndromes such as Alcardia syndrome, Cleidocranial dysostosis, foetal pyelectasis, gastroschisis, Gorlin syndrome, Jarcho-Levin syndrome, OEIS complex, VACTERL association and mucopolysaccharidosis.[5] Few anomalies of vertebral bodies not associated with neural tube defects have been reported in the literature.[6,7] Other terms used for this lesion are congenital scoliosis, unilateral aplasia of the body of the vertebrae, and complete unilateral failure of formation of the vertebral body.[8] Hemivertebra can equally be isolated or associated with defects of other major organ systems. We describe a case with clinical findings and radiological characteristics consistent

with spondylocostal dysostosis diagnosed at neonatal age with no associated anomaly.

Case Presentation

The patient was a 4-day-old boy, the fifth child of healthy, non-consanguineous parents who presented with swelling in the middle of the back. The mother was 30 years old, and the father was 58 at the time of the patient's birth. Family history was unremarkable, especially for congenital anomalies. The boy was born via spontaneous vertex delivery after a normal 40-week supervised gestation and no history of exposure to ionizing radiation during the pregnancy. His weight was 3,000 g, length 50 cm, occipitofrontal circumference 36 cm, and chest circumference 34 cm. There was a swelling at the mid portion of the back but there was no paucity of movement of any limb and no other gross dysmorphology. Other systems examinations were essentially normal (other than the musculoskeletal system). The results of examinations of blood and urine were all normal. Trunk radiographs showed thoracic spine scoliosis with convexity to the left side, a peak at T10 vertebra with a Cobb angle of 45^0 (severe scoliosis) and a focal kyphosis peak at T9 vertebra (Figure 1). Wedge-shaped vertebrae on the left side were noted involving T9 and T11 vertebrae (hemivertebrae), sagittal cleft was also noted involving T8 and T10 vertebrae, which represent butterfly vertebrae and absence of the right 9^{th} and 11^{th} ribs was also noted (Figure 1). However, their posterior elements and the remaining visualised vertebrae and ribs were normal. Computed tomographic (CT) scan of the trunk showed scoliosis at lower thoracic spine, focal kyphosis, hemivertebrae at T9 and T11, butterfly vertebrae at T8 and T10 and absence of 9^{th} and 10^{th} right ribs (Figure 2). It also showed a normal heart, great vessels, lungs and abdominal organs. The CT of the head showed normal brain. The diagnosis of isolated spondylocostal dysostosis in a neonate was made. The patient was clinically stable for three months of follow up at the time of writing this report.

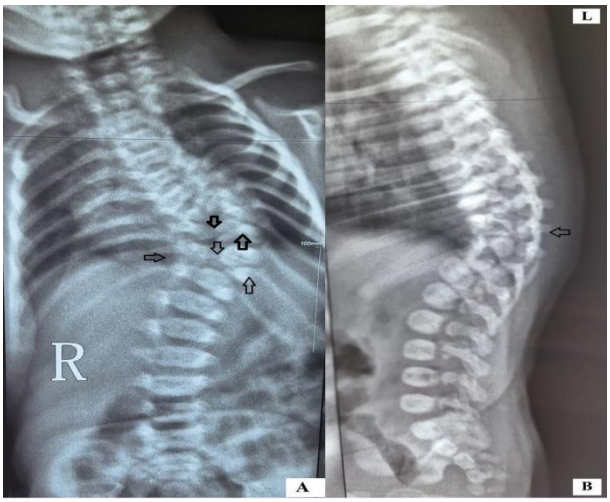

Figure 1: Anteroposterior (A) & lateral (B) views of trunk radiographs showing hemivertebrae of T9 & T11 (up arrows), butterfly vertebrae at T8 & T10 (down arrows), absence of right 11^{th} rib (left arrow), kyphosis peak at T9 vertebrae (right arrow) and scoliosis.

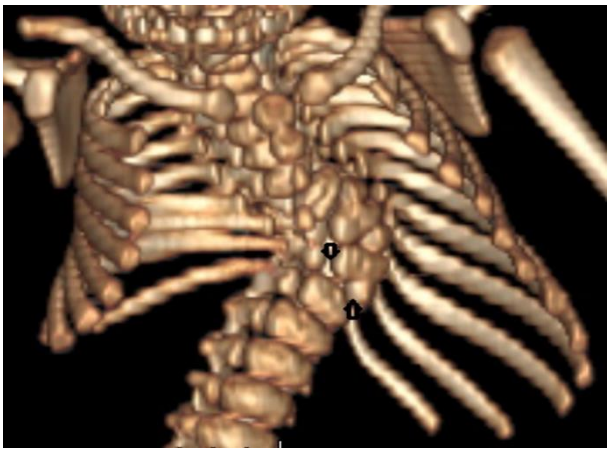

Figure 2: Volume rendered computed tomographic image of the trunk showing hemivertebra at T11 (up arrow), butterfly vertebra at T10 (down arrow), absence of right 9^{th} & 11^{th} ribs and Kyphoscoliosis.

Discussion

The first report of this distinctive complex of vertebral and rib malformations was that of Jarcho and Levin (1938), who described two siblings of the opposite gender with dysostosis of the vertebrae and associated rib anomalies.[9] Few cases in SCD patients were reported worldwide,[1,3] and also very few cases were reported in Africa (to the best of knowledge only three cases in literature).[3] The index case has no familial history of similar condition or teratogenic exposure which favours sporadic occurrence.

The affected baby was a male with multiple vertebral involvement. This contrasts with the described higher incidence for females compared to males. The spondylothoracic dysostosis (STD) is a strong differential of SCD.While STD is characterized with severe spinal and rib deformities which is associated with respiratory failure and high neonatal and early infant mortality, SCD has a wide variation of presentation and prognosis which some patients incidentally diagnosed as adult age. [1-3] However, this case has had no respiratory symptoms.

The cause of hemivertebra is unknown. One hypothesis has suggested that the hemivertebra may result from the intersegmental arteries of the vertebral column's abnormal distribution.[9,10] The distribution pattern of the anomaly does not implicate any specific environmental or genetic factor.

Hemivertebrae may be isolated or occur in multiple areas within the spine and are frequently associated with other congenital anomalies.[11-13] Multiple rib abnormalities including fused/broad ribs, absence and supernumerary ribs involving more than four ribs were reported in most cases.[1-3]

While our case had only the absence of two ribs. Genitourinary tract and cardiac anomalies are the most common extra-musculoskeletal anomalies seen. Index case had no cardiac or genitourinary tract involvement.

Anomalies of the gastrointestinal tract, renal, central nervous system and diaphragmatic and inguinal hernias have also been reported.[8] In contrast, the index case had none of these associations, hence diagnosed with a mild penetrance of this disorder. Approximately one-sixth of patients with vertebral body anomalies have associated malformations unrelated to the axial skeleton and spinal cord.[4,9]

Hemivertebra may be part of syndromes including Jarcho-Levin syndrome, Klippel-Fiel syndrome, VATER syndrome (vertebral anomalies, imperforate anus, tracheoesophageal fistula, and renal anomalies), VACTERL syndrome (VATER with cardiac and limb anomalies), OEIS (omphalocele, bladder extrophy, imperforate anus, and spine anomalies), the Potter sequence, and open spina bifida.[14]

When involving the thoracic spine, there is usually a missing rib on the affected side, presenting with an unequal number of ribs.

This is consistent with the findings in our subject. Failures of unilateral spine formation can be classified into segmented, semi-segmented, and non-segmented hemivertebra variants. The hemivertebra in our patient was the multiple, unilateral, segmented type involving the thoracic spine. Although the cause of most hemivertebrae is unknown and familial cases of idiopathic scoliosis have occasionally been reported, suggesting autosomal dominant inheritance.[15]

Since the spinal curve usually progresses more slowly in hemivertebrae than in idiopathic scoliosis, some familial members with mild scoliosis may have been overlooked. Such familial occurrence may be diagnosed by radiographic examination.

The prognosis of hemivertebra is related to the site of the affected vertebra, the number of affected vertebrae, and the associated anomalies. Thus, the prognosis is favourable in the index case. In addition, recent corrective surgery of congenital scoliosis caused by hemivertebra has much improved the prognosis. If no other major congenital malformations exist, the prognosis of hemivertebra may be favourable.

Conclusion

Spondylocostal dysostosis is a rare Musculo-skeletal abnormality involving the vertebrae and ribs, and presenting with a short trunk. A hemivertebra is a typical congenital vertebral anomaly with the potential for a severe spine deformity later in life (scoliosis).

Imaging plays a vital role in its diagnosis. An early diagnosis is necessary to anticipate the prognosis per the specific deformity to enable the stoppage by surgery. It is much better to do a relatively simple operation at a very young age to balance the growth of the spine than to wait until the deformity is severe when complex salvage procedures would be needed.

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Authors' contributions

FAG conceptualized the report, performed a literature search, and wrote the first draft. ML performed a literature search and reviewed the

manuscript. UHU searched the literature, prepared the images, and reviewed the manuscript. ZM analysed and interpreted the patient data regarding the images. All authors read and approved the final manuscript.

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Declarations

Ethics approval and consent to participate
Approval was obtained from the hospital research and ethics committee of the State Specialist Hospital Maiduguri (SSHM).

Consent for publication
Written informed consent was obtained from the patient's caregiver for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal or any other authorized person/body as appropriate.

Competing interests
The authors declare that they have no conflict of interest.

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