

Case Report

Jarcho-Levin syndrome term baby with recurrent apnea

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ABSTRACT

Jarcho-Levin syndrome or spondylocostal dysostosis, is a rare axial skeleton anomaly due to mutation in genes involved in somitic segmentation during embryogenesis. We describe a term, newborn baby with antenatally detected midthoracic lordotic deformity with skeletal features of multiple rib anomalies and vertebral defects, but with normal limb lengths. Baby had recurrent apnea requiring non-invasive ventilation and was successfully weaned to room air within 2 days. This syndrome is underdiagnosed and we wish to highlight the simple process involved in making a clinico-radiological diagnosis.

Keywords: Skeletal dysplasia, Spondylothoracic dysplasia, Jarcho Levin syndrome, Polythelia, Hemivertebra, Rib anomaly

INTRODUCTION

Jarcho-Levin syndrome is a rare skeletal dysplasia affecting the axial skeleton, occurring with an incidence of 1 in 40,000 live births.^{1,2} Inherited usually as an autosomal recessive trait by mutation in *DLL3* gene, sporadic occurrences are also reported rarely in literature.³ Here we describe a neonate who presented to us with recurrent apnea and characteristic skeletal findings.

CASE REPORT

A full-term female newborn presented to our NICU with antenatally detected mid thoracic lordotic deformity. Born to non-consanguineous parents, with no family history of malformations, recurrent abortions or history of teratogen exposure, the cause for the neonate's skeletal dysplasia remained undetermined. The baby's mother was under oral beta blockers for hypertensive disease of pregnancy from second trimester. The baby was born via operative delivery with no requirement for resuscitation at birth. Baby had a

short neck, polythelia, short trunk, protuberant abdomen and a sacral tuft of hair (Figure 1). No limb length discrepancy or limb anomalies were noted.



Figure 1: Neonate with right sided polythelia. Short neck, short trunk and protuberant abdomen are also noted.

On second day of life, baby had recurrent apnea requiring 2 cycles of bag and mask ventilation. She had to be placed on non-invasive ventilation for apnea and was weaned off within next 2 days. Skeletal survey revealed a short cervical region, multiple thoracic and sacral hemivertebrae, few absent vertebrae, asymmetrical crab-like ribs with absent and fused ribs (Figure 2). Ultrasound of spine did not reveal any neural tube defects. Ultrasound

abdomen, cranium and 2D echo ruled out any associated anomalies.

The above findings were consistent with clinico-radiological diagnosis of Jarcho-Levin syndrome. Parents were offered genetic counselling and orthopaedic consult for the baby. Baby was discharged and is under regular follow-up.



Figure 2: Radiographs of the neonate multiple segmentation defects of vertebra appearing like tramline with bifid, broadened and irregular ribs similar to a crab like configuration.

DISCUSSION

Jarcho-Levin syndrome (JLS) also known as spondylocostal dysostosis (SCD) was first described by Jarcho and Levin in 1938 as constellation of hereditary vertebral malformations.² It is rare skeletal dysplasia affecting the axial skeleton with worldwide incidence of 1 in 40000 live births.¹ This condition has to be differentiated from spondylothoracic dysostosis (STD) which has similar fan-like configuration of ribs, but with a higher degree of respiratory embarrassment.³ The inheritance is autosomal recessive with several genes associated with NOTCH signalling pathway implicated, most commonly the DLL3 gene.³

Skeletal malformations predominantly affect the chest wall and spine, sparing the extremities. Multiple segmentation defects of the vertebrae are noted with at least 10 segments affected, in combination with abnormalities of the ribs, which are maligned, broadened, bifid, with a variable number of irregular intercostal rib fusions, and sometimes with a reduction in rib number, giving it a crab like configuration.^{1,4} Short neck with congenital scoliosis suggests SCD, but presence of cervicothoracic fusion with rigid neck point towards the differential diagnosis, STD.³

Clinically, the diagnosis of SCD is suggested by a short-webbed neck, short trunk, protuberant abdomen, and scoliosis. Brachycephaly, wide nasal bridge, upslant palpebral fissures, low posterior hairline, pectus carinatum, lordosis, protuberant abdomen with inguinal or umbilical hernia may also be present.⁴ Airway anomalies have also

been described in these infants, contributing to respiratory failure in these infants, as in our case.⁵

Cleft soft palate, polythelia, cryptorchidism, hydronephrosis with ureteral obstruction, double collecting system, bilobed bladder, absent external genitalia, anal and urethral atresia, uterus didelphys, cerebral polygyria, neural tube defects, atrial septal defects, single umbilical artery, talipes equinovarus are rare associated congenital anomalies with SCD.⁴

The major cause of morbidity and mortality in SCD is thoracic insufficiency leading to decreased lung volume and underdevelopment leading to recurrent pulmonary infections.⁶ Significant thoracic restriction occurs in approximately 60% of newborns, resulting in some type of respiratory distress. Treatment of SCD depends on the degree of thoracic insufficiency, concurrent pulmonary complications, presence of chest wall defects, and quality of life. The surgical treatments for thoracic insufficiency in SCD include vertical expandable prosthetic titanium ribs (VEPTR) and chest wall reconstruction with latissimus dorsi flap transfers or polypropylene mesh.⁷ Non-severe cases may survive to adulthood, without any requirement for surgical interventions.

CONCLUSION

JLS is a unique skeletal dysplasia involving only the axial skeleton and routine survey may miss the crucial external findings. Characteristic features of asymmetrical ribs and vertebral anomalies helps in clinico-radiological identification of this syndrome and needs to be differentiated from its ominous, close differential, STD.

These infants need life-long follow-up due to their higher predilection for pulmonary infections.

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