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Jarcho-Levin Syndrome – A Case Report

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Summary:

Jarcho- Levin Syndrome is an eponym that represents a spectrum of short trunk skeletal dysplasia with variable involvement of the vertebrae and ribs. Initialy considered to be lethal, it is now accepted as compatible with life in its milder presentations. Neural tube defect is a rare association in Jarcho-Levin Syndrome. Here we report a case of Jarcho-Levin Syndrome with meningomyelocele in a newborn.

Key words: Jarcho-Levin Syndrome, Spondylo- thoracic dysplasia; Spondylo- costal dysplasia; Short trunk dwarfism, Meningomyelocele.

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Introduction:

Jarcho and Levin described a disorder of vertebral and costal anomalies in 1938 as Jarcho- Levin Syndrome[1]. There are two subtypes. One is Spondylo-thoracic dysplasia (STD) which suggests predominant vertebral defects and another is Spondylo- costal dysplasia (SCD) that describes the variant with vertebral and costal anomalies[2].This disorder comprises short trunk, prominent occiput, crab like rib cage associated with multiple vertebral defects and ribs that flare in a fan like pattern. Short neck, low posterior hair line, protruded abdomen with normal limbs, pectus carinatum are other anomalies. Occasional abnormalities may be cleft palate, hydronephrosis with ureteral obstruction, anal atresia and neural tube defects (33%).The purpose of reporting this case is to bring to clinical understanding of this rare disorder and review the emerging current knowledge about it.

Case Report:

A female term newborn 8 hour old presented with a swelling on the lower part of the back since birth. Mother Roksana, 38 year old, had irregular antenatal check up with good antenatal health according to her. Prenatal ultrasound was not done. Baby was delivered normally and cried immediately after birth. Her weight was 3 kg. Occipitofrontal circumference was 36 cm, length was 46 cm. Her neck and trunk was short and pectus carinatum was obvious (Fig.1). Vital signs were normal though chest cage was abnormal. Reflexes were good. The swelling was over the lumbar region of the back, covered with skin, cystic in consistency measuring 1.5 cm, nontender. Lower limbs were of normal length with active movements. There was no problem with bowel and bladder.

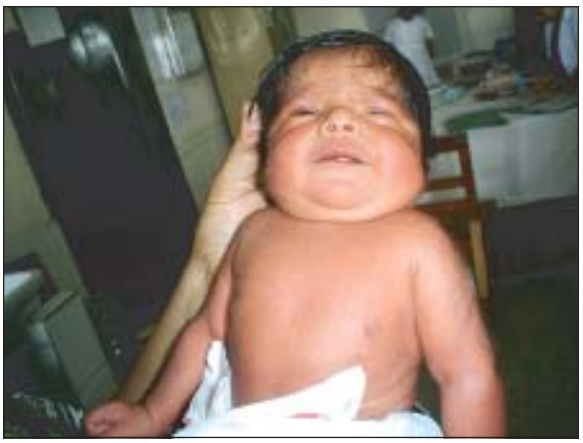

Fig.-1: *Showing short neck and pectus carinatum*

We consulted with the department of Neurosurgery of our medical college hospital regarding the swelling on the back; the neurosurgeon diagnosed this as meningomyelocele(Fig.2) and advised her to come for follow up visit after three months as the functions of the lower limbs were normal and there was no complain regarding bowel and bladder habits. The babygram revealed the classical radiological features of STD like pebbled beach appearance of the vertebrae and posterior fusion of the ribs, multiple hemivertebrae, fused vertebrae and abnormal rib cage (Fig. 3).

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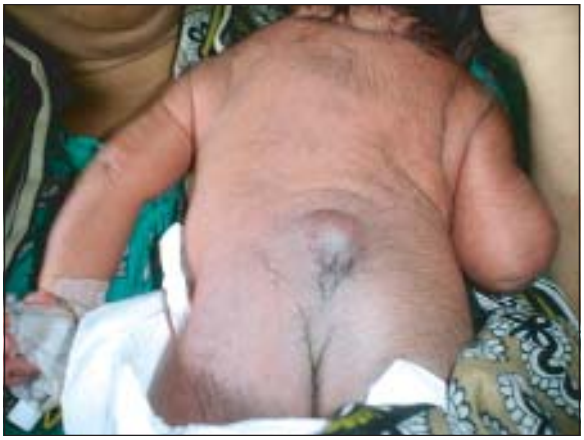

Fig.-2: *Showing meningomyelocele on the lumbosacral region of the patient*

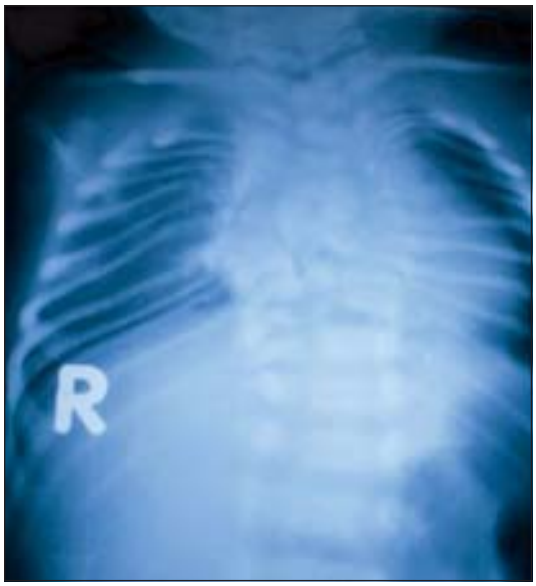

Fig.-3: *X-ray chest showing pebbled beach appearance of the vertebrae and posterior fusion of the ribs and multiple hemi vertebrae*

Finally we discharged the patient on request with the advice of consulting outpatient department of Orthopedics as soon as possible

Discussion:

Jarcho-Levin syndrome is a type of short trunk skeletal dysplasia with vertebral and rib anomalies. About 400 cases have been described in the world literature. It has both autosomal dominant and recessive modes of inheritance. The vertebral anomalies seen are hemi vertebrae, absent vertebrae, fused vertebrae, wedge vertebrae, sickle shaped vertebrae due to segmentation and formation defects [3-5]. (pebbled beach appearance). The costal defects noted are crab like or fan like appearances of the thorax due to crowded ribs, posterior fusion of the ribs and absent, irregular or bifid ribs.

This disorder has been noted in both consanguineous and non-consanguineous families. Most cases reported followed a pattern of autosomal recessive inheritance.

The case that we are presenting here has characteristic clinicoradiological features of Jarcho-levin syndrome with associated neurological abnormality like neural tube defect (Meningomyelocele). Neurological anomalies , such as neural tube defects and hydrocephalus are uncommon [6]. Solomon et al. [2] classified Jarcho-levin syndrome into 2 clinical phenotypes based on the extent and distribution of skeletal anomalies, the pattern of inheritance and the prognosis.

STD is an autosomal recessive disorder with posterior symmetric fusion of all the ribs at the costovertebral joints bilaterally and segmentation and formation defects of the vertebrae throughout the spine giving a classical 'crab like` or' fan like` appearance to the thorax. The ribs themselves had no defects. In STD neonatal death or death in infancy may occur due to pneumonias, restrictive lung disease.

SCD may be inherited in both autosomal dominant and recessive forms. Intrinsic rib anomalies like broadening, bifurcation and asymmetrical fusion are noted. The survival rate in SCD is high after the age of six months [7].Patients with SCD are known to have mutations in the delta-like 3 (DLL3) gene on chromosome 19.[8,9] .Patients with STD have no mutations in the DLL3 gene. Bannykh et al.[10] analysed protein expression from PAX1 and PAX9 genes in 2 sibs with this syndrome and compared it with age matched controls. Immunochemical analysis showed a significant reduction in the levels of protein expression on chondrocytes of the vertebral column.

Prenatal diagnosis by ultrasound can be done as early as 16 weeks of gestation after conception. Ultrasound criteria for diagnosis are unpaired or poorly formed vertebrae, indistinct or fused posterior ribs, irregular pebble like appearance of the spine, short trunk, protuberant abdomen, hernias and normal limb length[11].

Counseling the affected family is not a simple task because of varied presentation and striking intrafamilial variability. The exact clinicoradiological with molecular diagnosis is essential for accurate genetic counseling and prognostication of individual case.

Management should aim at aggressive neonatal care. Surgery of the spine to improve the thoracic volume and hence decrease the pulmonary restriction has been tried.

Conclusion:

Jarcho- Levin syndrome was considered initially lethal but it is now considered to be compatible with life in its milder presentation. So correct diagnosis and management can help both the affected newborn and the family by updated knowledge about this syndrome.

References:

1. Jarcho S, Levin PM. Hereditary malformation of the vertebral bodies. Bull Johns Hopkins Hosp 1938; 62: 216-26.
2. Solomon L, Jimenez RB, Reiner L. Spondylothoracic dysostosis: report of 2 cases and review of the literature. Arch Path Lab Med 1978; 102: 201-205.
3. Gellis SS, Feingold M. Spondylothoracic dysplasia (costovertebral dysplasia, Jarcho-Levin syndrome). Am J Dis Child 1976; 130: 513-14.
4. Pochaczevsky R, Ratner H, Perles D, Kassner G, Naysan P. Spondylothoracic dysplasia. Radiology 1971; 989(1): 53-58.
5. Pamela SK, Deborah D, Susan AB, Mary EMP. Jarcho-Levin syndrome: four new cases and classification of subtypes. Am J Med Genet 1991; 40; 264-70.
6. Giacoia GP, Say B. Spondylocostal dysplasia and neural tube defects. J Med Genet 1991; 28: 51-53.
7. Cornier AS, Ramirez N, Arroyo S, Acevedo J, Garcia L, Carlo S et al. Phenotype characterization and natural history of spondylothoracic dysplasia syndrome: a series of 27 new cases. Am J Med Genet 2004; 128A; 120-26.
8. Turnpenny PD, Bulman MP, Frayling TM, Abu- Nasra TK, Garrett C, Hattersley AT et al. A gene for autosomal recessive spondylocostal dysostosis maps to 19q13.1-q13.3. Am J Hum Genet 1999; 65: 175-82.
9. Bulman MP, Kusumi K, Frayling TM, Mc Keown C, Garrett C, Lander ES et al. Mutations in the human delta homologue, DLL3, cause axial skeletal defects in spondylocostal dysostosis. Nature Genet 2000; 24: 438-441.
10. Bannykh SI, Emery SC, Gerber JK, Jones KL, Benirschke K, Masliah E. Aberrant PAX 1 and PAX 9 expression in Jarcho-Levin syndrome: report of two Caucasian siblings and literature review. Am J Med Genet 2003; 120A: 241-246.
11. Whittock NV, Sparrow DB, Wouters MA, Silience D, Ellard S, Dunwoodie SL et al. Mutated MESP2 causes spondylocostal dysostosis in humans. Am J Hum Genet 2004; 74:1249-1254.