

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. Initially considered to be lethal, it is now accepted as compatible with life in its milder presentations. Neural tube defect is a rare association

Introduction:

Jarcho and Levin described a disorder of vertebral and costal anomalies in 1938 as Jarcho- Levin Syndrome¹. 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 anomalies². 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 Rokhana, 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

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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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 defects³⁻⁵. (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 uncommon⁶. 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 months⁷. 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 length¹¹.

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.

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