

Spondylocostal dysplasia and neural tube defects

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Abstract

Spondylocostal dysplasia (Jarcho-Levin syndrome) comprises multiple malformations of the vertebrae and ribs coupled with a characteristic clinical picture of short neck, scoliosis, short trunk, and deformity of the rib cage. We describe a patient with the syndrome who also had spina bifida and diastematomyelia. We surmise that this association is not coincidental. Additional evidence is needed to support the hypothesis that spondylocostal dysplasia and neural tube defects are aetiologically related.

Spondylocostal dysplasia is a congenital disorder with multiple abnormalities of the vertebrae and thoracic

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cage resulting in short trunked dwarfism. It has been variously reported as Jarcho-Levin syndrome, 'hereditary multiple hemivertebrae', 'bizarre vertebral anomalies', 'costovertebral dysplasia', and Covesdem syndrome when associated with mesomelic shortening of the limbs. Despite the major vertebral segmentation defects, including spina bifida occulta, spondylocostal dysplasia is considered unrelated to neural tube defects.¹

The purpose of this paper is to present a case of spondylocostal dysplasia associated with spina bifida and diastematomyelia and to review the pertinent published reports.

Case report

The proband, a male, was born to a 28 year old woman after a 38 week, uncomplicated pregnancy. Apgar scores were 4 and 7 at one and five minutes, respectively. Shortly after birth, the infant had considerable respiratory difficulty requiring intubation in the delivery room and subsequently neonatal intensive care. The thorax was grossly deformed with shortening of the trunk and neck and a protuberant abdomen. The occipitofrontal head circumference



Figure 1 Appearance at birth showing diastematomyelia and open sac of meningocele.

was 36.2 cm (>90th centile). There was a triangular opening of the mouth with cleft palate and brevicollis. The spine was short and exhibited marked thoracolumbar lordosis and dextroconvex scoliosis. Spina bifida cystica was present in the thoracolumbar area and diastematomyelia (diplomylelia) was noticed in the defect (fig 1). There was an imperforate anus associated with a perineal fistula. The genitalia were normal except for undescended testes. His extremities were well developed but showed paralysis of the legs. The remainder of the physical examination was within normal limits. CT scan of the head showed moderate hydrocephalus owing to aqueductal stenosis. An abdominal sonogram showed normal kidneys. X ray of the spine showed vertebral anomalies at all levels. Extensive and severe developmental anomalies of the vertebral bodies and widely open and spread neural arches were most marked in the lower cervical spine and in the thoracolumbar area. Hemivertebrae and block vertebrae were present, while other vertebral bodies were partially fused and irregularly deformed. The sacrum was least involved. The rib cage exhibited bizarre features with ribs of different size, thickness, and orientation (fig 2). Fused ribs were also present. An atrial septal defect was detected by two dimensional echocardiography. Chromosome studies using high resolution banding gave normal results (46,XY). The infant has remained dependent on a respirator and had two episodes of pneumonia. He underwent repair of the meningomyelocele and placement of a ventriculoperitoneal shunt. The infant's parents are

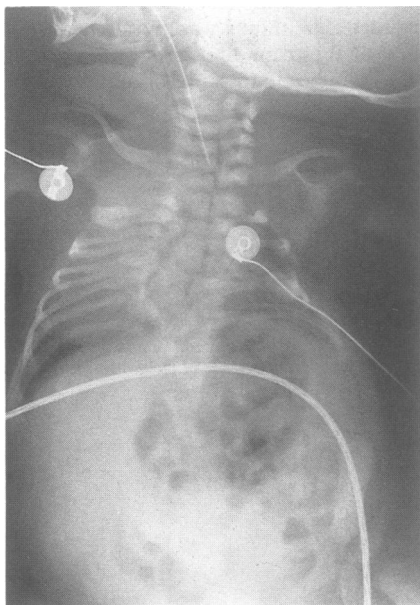


Figure 2 Anteroposterior radiograph showing shortening of the spine and vertebral and rib abnormalities.

both American Indian, remain unmarried, and are non-consanguineous. The mother denies knowledge of similar cases in their respective families. There was no history of teratogen exposure during the pregnancy. The father is 30 years old. A 5 year old sib is normal.

Discussion

In 1975, Wynne-Davies² made the observation that sibs of patients with localised multiple vertebral anomalies had an increased incidence of anencephaly or spina bifida cystica compared to the incidence in the general population and suggested an aetiological link between these two conditions. Subsequently, Naik *et al*³ performed a radiological necropsy study of vertebral and rib malformations in children with myelomeningocele. Sixty-four had anomalies, including fused vertebral arches, fused vertebral bodies, absence of vertebral bodies, and absent or fused ribs. Similarly, anomalous ribs seem to occur with increased frequency in patients with myelodysplasia. In the series reported by McLennan,⁴ the first nine ribs were abnormal and vertebral anomalies were found concomitantly with abnormal ribs in about 50% of patients with spina bifida.

Lendon *et al*⁵ compared the radiological records of patients with multiple vertebral anomalies and a comparable number of patients with spina bifida cystica. The incidence of hemivertebrae and of rib, vertebral body, and vertebral arch fusions were determined. The total number of bone anomalies in the multiple vertebral anomalies group far exceeded that seen in the spina bifida cystica group; however, the distribution of these anomalies were similar in both groups. Although the study of Lendon *et al*⁵ suggests an aetiological connection, it does not provide proof of such an association.

Spina bifida occulta appears to be a common finding in spondylocostal dysplasia (Jarcho-Levin syndrome). We surveyed 59 reported cases with this condition and found spina bifida occulta in 24 (40.6%).^{1 6-9}

Until recently, neurological abnormalities were not considered to be associated with the Jarcho-Levin syndrome. Poor *et al*⁷ described a case of this syndrome associated with cerebral polygyria. Reyes *et al*⁸ reported a case of Jarcho-Levin syndrome associated with diastematomyelia. These authors claim that neurological abnormalities should be considered a component of the Jarcho-Levin syndrome. This suggestion is further supported by the review of a case published in 1976 in which thoracolumbar rachischisis accompanied the cardinal features of the Jarcho-Levin syndrome.⁹ The infant was born to a woman who abused lysergic acid diethylamide during pregnancy. The case reported here is unique in that diastomyelia and meningomyelocele occurred in association with findings characteristic of the Jarcho-

Levin syndrome. Pathological reports indicate that at necropsy partial or complete diastematomyelia occurs in about 30 to 35% of spinal cords involved in meningomyelocele.¹⁰

Our case also presented non-skeletal malformations previously described in patients with Jarcho-Levin syndrome.⁷ They included cleft palate, triangular opening of the mouth, imperforate anus, undescended testes, and dextroposition of the heart with atrial septal defect. Macrocephaly was the result of hydrocephalus accompanying aqueductal stenosis, a feature not previously described.

The Jarcho-Levin syndrome is commonly familial but sporadic cases are well recognised. In a recent review of 39 patients with this syndrome, 35 were considered as having a recessive disorder and four a dominant one.¹

It is possible that the Jarcho-Levin syndrome is under-reported. The concomitant findings of neural tube defects may preclude its recognition as a separate entity because of the well known association of vertebral and rib anomalies with spinal dysraphism. The involvement of vertebral bodies at all levels of the spine is an important diagnostic feature in the Jarcho-Levin syndrome.

The full phenotypic manifestations of a rare autosomal recessive gene become evident only when a sizable number of affected patients have been evaluated. Although a putative association between the Jarcho-Levin syndrome and malformations of the

spinal cord remains contentious, study of familial cases of the syndrome may provide evidence of an aetiological link. The concomitant presence of spondylocostal dysplasia in some members of a family and neural tube defects in others would prove that the relationship between the two conditions is not merely coincidental. The genetic implications of the diagnosis of the Jarcho-Levin syndrome highlight the importance of its recognition.

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