Spondylocostal dysplasia and neural tube defects

George P Giacoia, Burhan Say

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...

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 brevicolli. The spine was short and exhibited marked thoraco-
lumbar lordosis and dextroconvex scoliosis. Spina
bifida cystica was present in the thoracolumbar area
and diastematomyelia (diplomyelia) 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 dimen-
sional 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 meningomyelecele and placement of a
ventriculoperitoneal shunt. The infant’s parents are
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 sibling is normal.

Discussion
In 1975, Wynne-Davies\(^2\) 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, includ-
ing 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 con-
comitantly 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 con-
sidered 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
diastematomyelia and meningomyelocoele occurred in
association with findings characteristic of the Jarcho-

![Figure 2: Anteroposterior radiograph showing shortening of the
spine and vertebral and rib abnormalities.](image-url)
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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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G P Giacoia and B Say

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