

Spondylocostal Dysostosis (Jarcho-Levin Syndrome) In An Arab Family

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There are two distinct forms of congenital spinal deformities involving the entire vertebral column with short trunk dwarfism and rib anomalies namely severe and mild forms of Jarcho-Levin syndrome. We present two new related cases that we believe are the first reported cases among the Arab population. They are examples of the recessive mild form of the syndrome affecting the entire spine including cervical and sacral regions with multiple rib fusion. On advising parents regarding the prognosis caution must be paid as many patients show good tolerance of their deformities.

Bahrain Med Bull 1999;21(1): 30-32

Hereditary costovertebral malformation associated with short trunk dwarfism was first described by Jarcho and Levin in 1938¹. Since then, other investigators reported different forms of the condition under various terms. These were listed by the MIM entries 122600, 271520 and 277300.

The racial distribution of all reported cases show predominance of Hispanic families². Recently, more cases were reported among Caucasian, white and black races^{3,4}. From Asia only one Japanese and one Chinese cases were reported^{5,6}.

We present two new related cases that we believe are the first reported cases among the Arab population to be added to reported cases in the literature in a trial to accumulate more information that will aid the discrimination between the different forms of the syndrome and help in identification of the overall prognosis.

THE CASES

We report a brother and a sister, 4.5 and 7 years old. They are the second and third among the other two normal siblings. Their parents are second degree cousins.

The girl presented in our hospital for the first time at the age of 15 months with bilateral inguinal hernia. History of normal pregnancy and delivery and low birth weight (not specified), however the length at birth was not identified. At that time multiple vertebral and rib deformities were observed together with short stature. After reviewing the

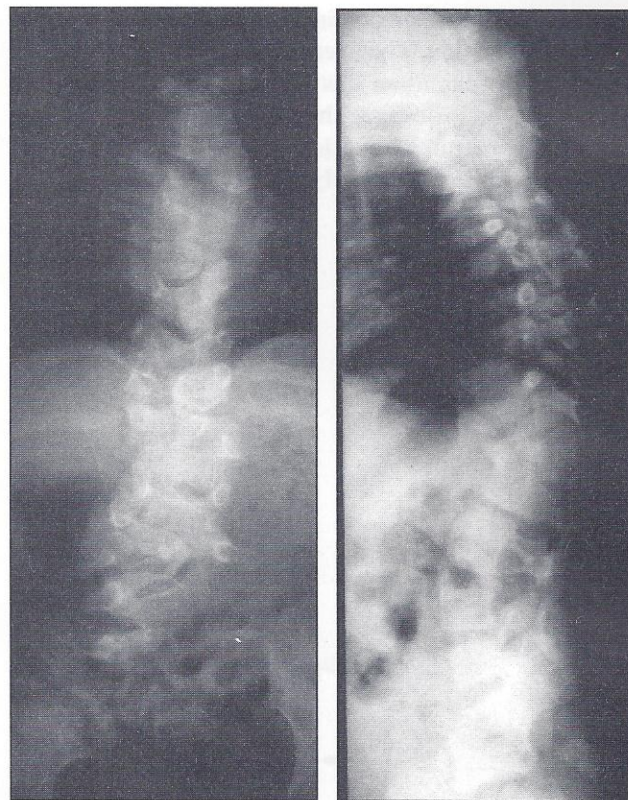


Figure 1. Radiographs of the female patient. AP and lateral views at latest follow up visit showing multiple deformities of all thoracic and lumbar vertebrae with multiple rib fusions.

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literature diagnosis was made as mild autosomal recessive form of spondylocostal dysostosis. General examination of the girl revealed prominent occiput, broad forehead, anteverted nares, short mobile neck, short barrel shaped and asymmetric chest with scoliotic spinal curve, bilateral inguinal hernia, diastasis of recti and long thin limbs. Radiological examination showed multiple extensive vertebral anomalies especially hemi-vertebrae, butterfly vertebrae and spina bifida involving the whole spine from cervical down to the sacral region. There were absent transverse processes in most of the lumbar vertebrae with widening of spinal canal. Multiple rib fusion was also noted. Figure 1 shows radiographic pictures for the girl at time of presentation and latest follow up visit.

Shortly after his birth the brother was assessed and found to have the same syndrome with manifestations similar to his sister with some differences including that he has wide nasal bridge, his neck is fixed and there is spinal lordotic curve together with scoliosis. He had an umbilical hernia. Radiological examination of the boy showed the same anomalies seen in his sister with the same extension. Rib abnormalities are less in the boy than in his sister. Figure 2 shows radiographic pictures for the boy at time of presentation and latest follow up visit.

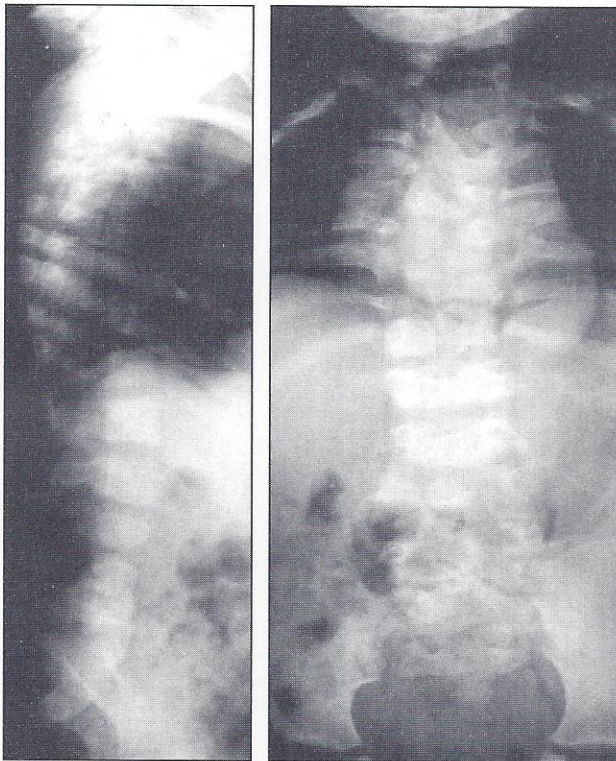


Figure 2. Radiographs of the male patient. AP and lateral views at latest follow up visit showing same deformities with less prominent rib anomalies.

Patients were assessed every six months and it was noted that both are of normal intelligence and they have no neurological deficits.

DISCUSSION

Two distinct forms of Jarcho-Levin syndrome have been identified; the severe and mild forms⁷. It was not before

1986 when Ayme and Preus proposed a comprehensive classification of the condition according to the mode and pattern of inheritance, the severity of costovertebral malformation and the presence of associated visceral anomalies. They found that the severe form is always autosomal recessive while the mild form showed 2 genotypes, autosomal dominant and autosomal recessive⁸.

Severe form is characterized by crab-like deformities of the ribs causing still-birth or death in early infancy due to respiratory failure. In the mild form the ribs are slightly affected, the short trunk dwarfism is a standard feature and patients have a normal life expectancy^{7,9}.

The condition in its two forms is entirely different from congenital scoliosis, although both are associated with particular groups of malformation². In congenital scoliosis only limited segments of vertebral column are affected with normal thoracic cage and absent disproportionate dwarfism. In Jarcho-Levin syndrome, all vertebrae are affected, although the cervical spine may be normal or partially involved. Scoliosis is seldom severe in both forms of the syndrome¹⁰.

Non skeletal involvement is well established. Congenital heart diseases and renal anomalies are the commonest. Urogenital abnormalities, polydactyly, tracheo-oesophageal fistulae and anal atresia have also been reported^{2,11}. The non-skeletal anomalies in our cases were also reported by few authors^{2,7}.

Several characteristic features of the disease were described by various authors including prominent occiput, macrocephaly, broad forehead, upslanted fissures, wide nasal bridge, anteverted nares, prominent philtrum, triangular mouth, protruding tongue, cleft palate, short fixed neck and barrel shaped chest¹². Most of these features were shown in our cases.

Our cases are still young, but their good general condition confirm the hypothesis of normal life expectancy in the mild form^{8,13}.

Ultrasonography is mentioned to have a valuable antenatal diagnostic role for the condition¹⁴. Some authors could differentiate between the two forms by ultrasound and their findings were confirmed postnatally. At 23 weeks of gestation they visualized a shortened spine, disorganization of vertebral bodies, posterior fusion of the ribs and normal long-bone biometry¹⁵. The prenatal diagnosis of Jarcho-Levin syndrome allows aggressive neonatal care of the affected child, who may develop respiratory failure soon after birth. This will lead to an excellent outcome and improve survival¹⁴. On the other hand termination of pregnancy was elected by some authors after prenatal sonographic diagnosis of the disease¹⁵.

Familiarity with the syndrome in its various forms is necessary in order to avoid an unjustifiably pessimistic prognosis and to permit correct parent counseling. The prognosis can be considered good if the chest is not severely malformed. However, one must be cautious in advising families of the prognosis for a child with severe structural chest deformity, since it may not be severe from a functional point of view and such patients have demonstrated very good tolerance of their deformities. This is further helped

by normal or near normal central nervous system function.

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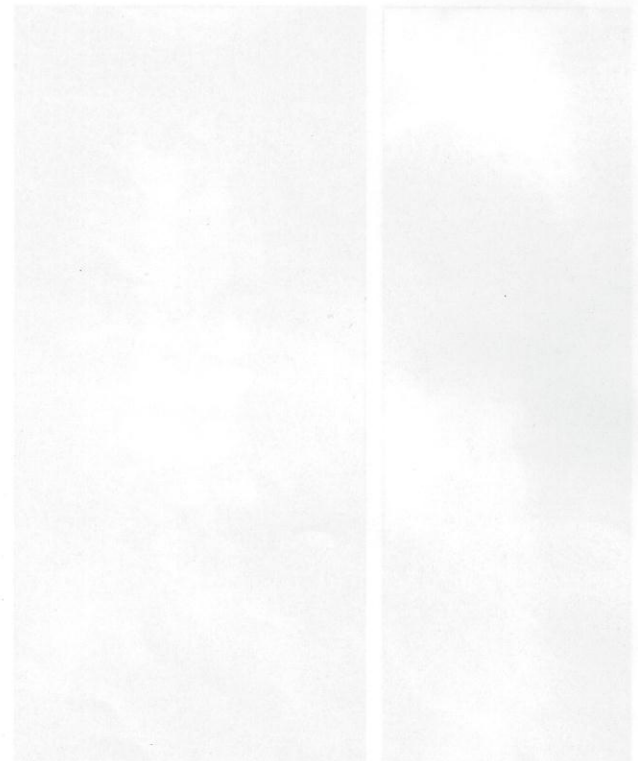


Figure 2. Radiographs of the spine patient of Jarcho-Levin syndrome. The left panel shows a normal spine and the right panel shows a spine with Jarcho-Levin syndrome. The vertebral bodies are small and the intervertebral spaces are narrow. The spine is curved and the patient has a hunched posture.