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Spondylocostal Dysostosis

Thirteen New Cases Treated by Conservative and Surgical Means

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Study Design. Prospective assessment of a cohort of patients affected by spondylocostal dysostosis.

Objective. To report on the results of conservative and operative management of spondylocostal dysostosis and, based on this, to propose an assessment and treatment protocol for the condition.

Summary of Background Data. Spondylocostal dysostosis and spondylothoracic dysostosis are subtypes of Jarcho-Levin syndrome, a hereditary condition manifested by vertebral body and related rib malformations. Mortality prevails in spondylothoracic dysostosis because of more severe respiratory compromise.

Methods. Details of prenatal and postnatal diagnosis, history, and management of 13 patients with spondylocostal dysostosis are presented. All patients were treated postnatally with repeated chest physiotherapy. Two patients refractory to conservative treatment underwent surgical intervention: the first had a chest wall reconstruction *via* a latissimus dorsi flap, the second a posterior spinal instrumented fusion for progressive scoliosis.

Results. Prenatal ultrasound in 4 of 13 cases showed full details of vertebral and rib anomalies. Thoracic and lumbar hemivertebrae were most common, leading to congenital scoliosis in 10 of 13 cases. A number of extraskeletal abnormalities were also identified. At an average follow-up of 4.5 years, the survival rate was 100% with a remarkable decrease of the rate of respiratory complications. Surgical treatment in selected cases led to satisfactory results.

Conclusions. Prenatal diagnosis of spondylocostal dysostosis allows exclusion of spondylothoracic dysostosis and aids genetic counseling in quantifying the risk to siblings. Postnatally, prompt management of these patients with physiotherapy leads to prolonged survival. Surgical intervention may then be indicated to stabilize chest wall or spine deformities, with promising results.

Key words: congenital scoliosis, spondylocostal dysostosis, operative treatment. **Spine 2004;29:1447–1451**

In 1938, Jarcho and Levin described a syndrome affecting individuals with short neck and short-trunk dwarfism resulting from multiple vertebral and associated rib anomalies. In Jarcho-Levin syndrome (JLS), hemivertebrae, butterfly vertebrae, and unsegmented, hypoplastic vertebrae may almost affect the entire thoracic and lumbar spine at alternate levels.[1] Cervical spine involvement may be in the form of hemi- and unsegmented vertebrae, causing shortness of neck. The ribs can vary in number and shape often showing fusion, primarily near the costovertebral ends. Multiple rib and vertebral anomalies may determine scoliosis, kyphosis, and pectus deformities. The widespread distribution of the vertebral anomalies and their association with rib anomalies differentiate JLS from congenital scoliosis.[1] In 1978, Solomon *et al* classified cases of JLS into two types: spondylothoracic dysotosis (STD) and spondylocostal dysotosis (SCD).[2]

Individuals with STD have vertebral anomalies and severely deformed and fused ribs, resulting in the "crab-like" appearance of the chest on plain radiographs. The inheritance pattern is autosomal dominant. Patients affected by STD have a higher fatality rate as a result of posterior tethering of the ribs, leading to progressive thoracic restriction with growth.

Individuals with SCD have vertebral anomalies and intrinsic rib defects or malformations (hypoplasia) of varied patterns. The inheritance pattern is either autosomal dominant or recessive, the latter being described as the most aggressive form. Patients with SCD survive more often than those with STD despite gross thoracic abnormalities, possibly because the lungs are not restricted. SCD has a reported prevalence of $0.25/10^5$.[3]

The aim of this study is to outline a protocol for the assessment and treatment of SCD based on the results of conservative and surgical treatment in 13 new cases of this syndrome and the pertinent literature. We also describe the results of surgical treatment in 2 of our patients, including the use of the latissimus dorsi flap for costal defect reconstruction in one and instrumented posterior spinal fusion in the other.

To the best of our knowledge, the present series is the largest ever reported on the condition. From 1938 to date, 41 cases of SCD and 20 cases of STD have been described in the English medical literature, the largest single series reporting on no more than 5 and 6 patients, respectively.[2]

Materials and Methods

We performed a prospective follow-up study of 13 pediatric patients affected by SCD and treated at two Pediatric Orthopedic Institutions.

Five patients had consanguineous parents. Four patients were seen from birth and prenatal ultrasound diagnosis had been made in all of them. All patients had full spine and chest plain radiographs; six had ultrasound[4] evaluation of segmental defects and three had full spine MRI scans. A thorough clinical and radiologic assessment was done in all cases for cardiovascular,[5] genitourinary, parenchymal organ defects and neural tube defects.[6]

Our protocol in managing these patients postnatally was based on chest physiotherapy (CPT) and regular clinical eval-

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Table 1. Patients' Details

Case No.	Age First Seen	Sex	Consanguinity	Prenatal Diagnosis	Vertebral Defect Site	Associate Anomalies	No. of Absent Ribs		Rib Fusion	Rate of LRTI per Month	
							R	L		Pre-Rx	Post-Rx
1	Newborn	M	+	+	TL	ASD	5	0	−	3	1:3–6
2	Newborn	F	−	−	T	Jaundice, left renal agenesis, clubfeet	4	0	−	3–4	1:3
3	6 mo	F	+	−	T + L	Hydrocephalus, spina bifida, bifid uvula, supemumerary nipple	1	5	7,8,9 (L) 4,5 (R)	5–6	1:2
4	3 mo	M	−	−	TL	Sprengel's deformity	3	0	−	1–2	1:4
5	2 mo	F	−	+	T	Polycystic kydney	0	2	−	3	1:3
6	5 mo	F	+	−	T	−	2	0	5,6,7 (R)	4–5	1
7	5 mo	M	−	−	CT + TL	ASD, supemumerary nipple	4	0	6,7 (R)	3–4	1:3
8	Newborn	M	−	+	TL	ASD, diaphragmatic hernia	0	4	−	1	1:8
9	2 mo	M	−	−	T	Hydrocephalus, spina bifida, clubfeet	0	3	−	4–5	1
10	Newborn	F	+	−	TL	VSD, left renal agenesis	4	0	−	3–4	1:3
11	1 mo	M	+	+	T	Cleft palate, supemumerary nipple, DDH	2	3	−	4	1:3
12	3 mo	F	−	−	CT + TL	Jaundice, clubfeet	4	0	−	2–3	1:3
13	10 yr	M	−	−	T	−	3	3	6,7 (R)	4	1:2

CT = cervicothoracic; T = thoracic; TL = thoracolumbar; L = lumbar; VD = vertebral defect; ASD = atrial septal defect; VSD = ventricular septal defect; DDH = developmental dysplasia of the hip; LRTI = lower respiratory tract infection.

uations. Chartered physiotherapists treated patients on a daily basis in hospital and at weekly intervals after discharge, until improved clinical conditions allowed monthly appointments. Treatment consisted of a modified CPT routine followed by breathing games to perform diaphragmatic breathing in infants, and of positive expiratory pressure therapy plus chest wall oscillation in children.

The frequency of lower respiratory tract infections (LRTIs) was recorded. In the clinical setting of an LRTI, *i.e.*, worsening general conditions, oxygen desaturation, raised white cell count, and positive chest radiograph, patients were admitted to hospital to receive broad-spectrum intravenous antibiotic coverage. Antibiotics were then targeted to sputum culture results and discontinued at the reversal of the above clinical indexes.

Bracing was used in those cases warranting treatment of a progressive spinal deformity, provided it was tolerated by patients and on the grounds of a satisfactory pulmonary function.

A rib defect reconstruction procedure was performed in patient 8 (Table 1) who had frequently recurrent LRTIs, paradoxic chest wall movements, and a diaphragmatic hernia. Ultrasound showed herniation of the lower left lung lobe with the diaphragm and the upper pole of the spleen with respiratory movements. The rib defect was repaired with an autologous, vascularized latissimus dorsi flap to stabilize the chest wall and contain the lung and spleen herniation.[7]

Posterior instrumented spinal fusion was done in patient 13 (Table 1)[2,8] who was referred to our service at 10 years of age for progressive scoliosis. At the time of the first observation, he already had features of lower thoracic paraplegia,[2] restrictive lung disease, and recurrent and severe chest infections. His sitting balance had been deteriorating because of increasing spinal deformity and pelvic obliquity. Figure 1 shows his preoperative plain radiographs. Because of his poor general health, posterior instrumented spinal fusion with limitation of fusion levels from the lower thoracic vertebrae to the pelvis was the only management option at that time. Figure 2 shows his 3-year follow-up films, with adding-on of the upper thoracic curve and satisfactory maintenance of the lower spinal and pelvic deformity. At this time, his general health had improved to the stage that proximal extension of the spinal fusion was deemed feasible.

■ Results

There were 7 males and 6 females. Patient age varied from 1 day to 10 years when first seen. Final follow-up ranged from 26 to 72 months, averaging 56 months. No patient was lost to follow-up.

The nature of the vertebral defects was delineated in all cases. One or more vertebral anomalies were present in all patients and included all together 1 hypoplastic sacral vertebra, 3 absent thoracic vertebrae, 7 butterfly vertebrae (4 thoracic, 3 lumbar), and 18 hemivertebrae. Hemivertebrae, the most common vertebral anomalies, were more commonly located in the thoracic and thoracolumbar region than in the cervical region. Congenital scoliosis was identified in 10 of 13 patients (75%): average anteroposterior Cobb angles varied from 23° at the beginning of the observation to 38° at the latest follow-up.

Both unilateral (10 cases) and bilateral (3 cases) rib defects were noted. Rib fusion was found in 3 cases. Rib anomalies included posterior fusion, absence, and irregular size and shape.[9]

The extraskeletal associated anomalies[10] included supernumerary nipples (3 cases), clubfoot deformity (3

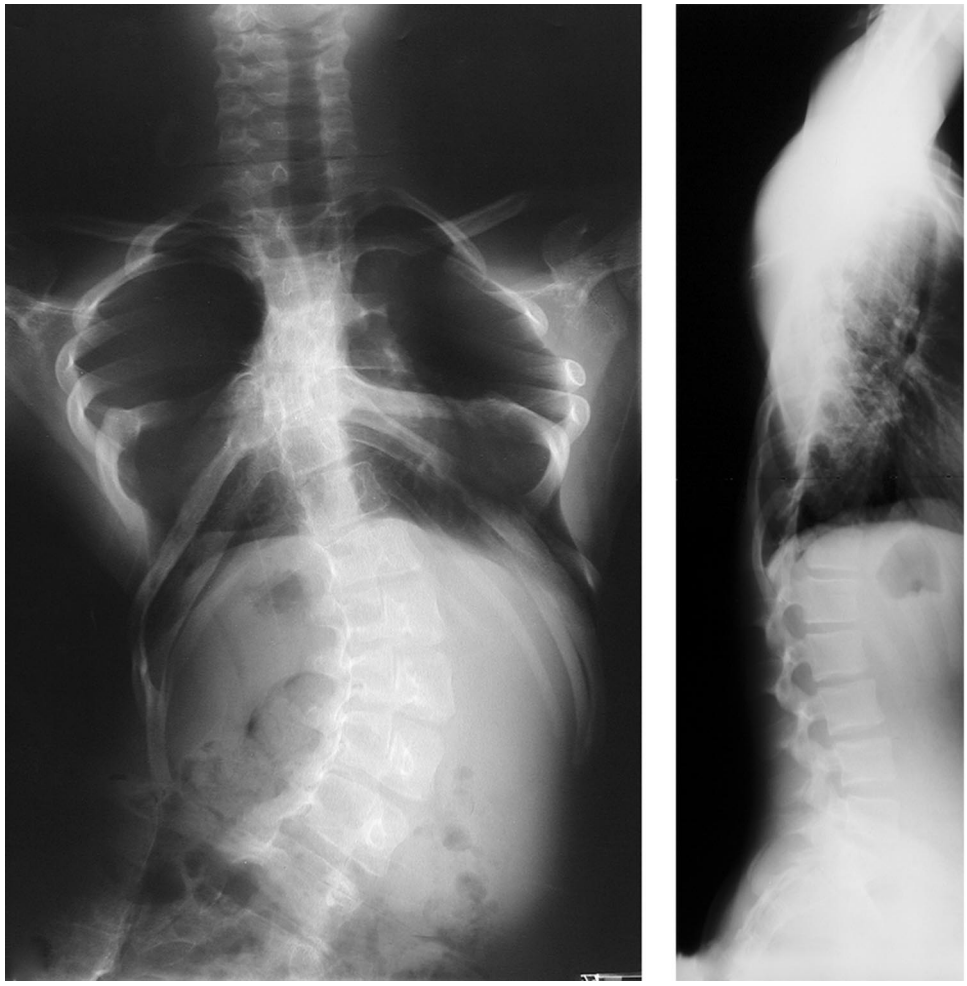

Figure 1. Case No. 13 preoperatively, aged 10 years. PA + lateral sitting films.

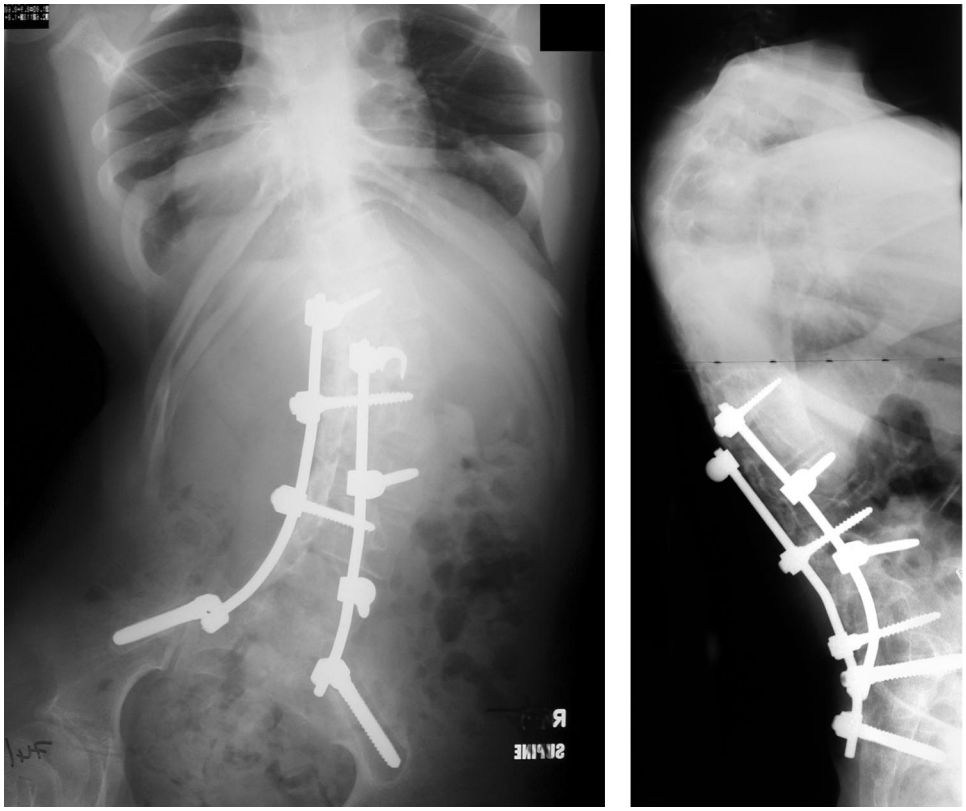

Figure 2. Case No. 13 2 years after posterior spinal fusion, aged 13 years. PA + lateral sitting films.

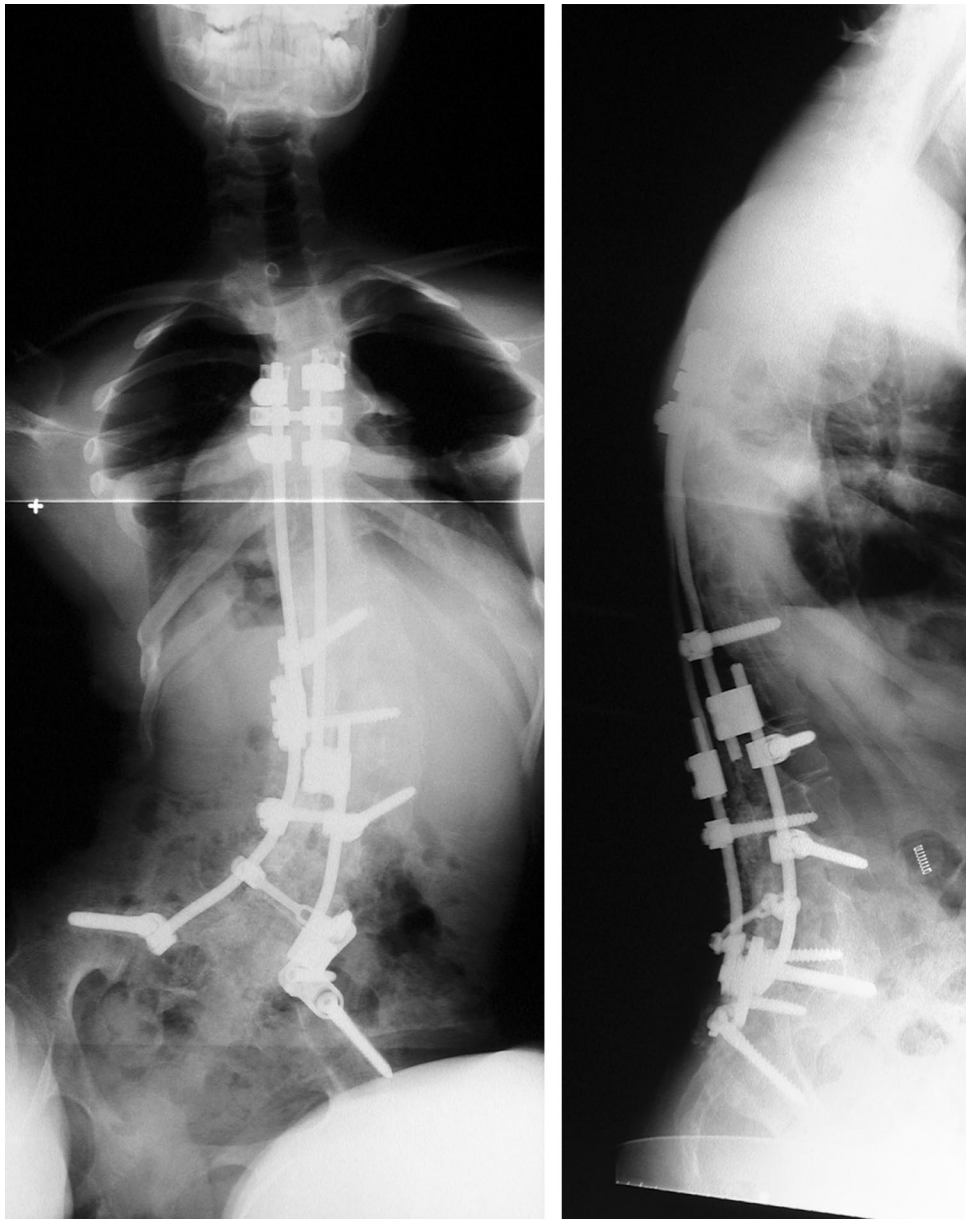

Figure 3. Case No. 13 2 years after thoracic extension of spinal fusion, aged 15 years. PA + lateral sitting films.

cases), Sprengel's deformity (1 case), strabismus (1 case), bifid uvula (1 case), cleft palate (1 case), congenitally dislocated hip (1 case), neonatal jaundice (2 cases), myelomeningocele and hydrocephalus (2 cases), renal anomalies (4 cases), atrial septal defect (1 case), and ventricular septal defect (1 case).

The presenting complaints in all patients were recurrent LRTIs, with an average of 3 episodes per month (range, 1–6), poor general condition, and failure to thrive. After birth, patients were hospitalized on average once a month (range, 0.3–4) mostly for treatment of LRTIs. Mechanical respiratory support was necessary once every three admissions to hospital for an average of 2 days (range, 1–5) during hospitalization. Mean height and weight growth curves were under the fifth and around the 10th centiles, respectively, before treatment.

This picture was reversed at follow-up, with a reduction of the incidence of LRTIs to an average of 1 episode every 3 months (range, 1:8 to 1 a month) and hospitalization to an average of once every 6 months (range, 1:8 to 1:3 months). We observed a mean reestablishment of the weight curves to the 25th centile and fairly stable height curves just under the fifth centile.

Patient 8, treated with autologous rotation latissimus dorsi muscle flap, at follow-up reveals containment of herniation, a supple scar, and a static rib defect. His pulmonary function improved readily after surgery[7] and currently, 24 months after surgical intervention, he experiences 1 episode of respiratory infection every 8 months. His paradoxical chest movements have stopped and his diaphragmatic hernia is well contained.

The follow-up for patient 13 is now 2 years following the revision procedure. Currently, the patient is pleased by his sitting balance and by the absence of uneven pressure areas or spinal instrumentation prominence. Although he his still frequently admitted to our Hospital

for respiratory care, the rate of his chest infections has fallen from 4 a month to 1 a month. Figure 3 shows satisfactory fusion and absence of instrumentation failure in his latest follow-up spine radiographs.

■ Discussion

SCD is a rare[3] disorder of vertebral and rib formation of unknown etiology. In our series, there was history of maternal diabetes in one of the patients only, confirming previous reports of low incidence of diabetic embryopathy in JLS.[3,8] Regarding its pathogenesis, the vertebral anomalies observed in this syndrome could be secondary to a developmental disturbance at the fourth to eight week of fetal life. At that time, the multiple centers of chondrification formed about the notochord unite to form a complete cartilaginous pro-vertebra composed of centra in the vertebral body and transverse, articular, and spinous processes. Failure of these chondrification centers to develop or unite could result in the development of hemivertebrae and butterfly vertebrae. The anomalous rib development is apparently secondary to the vertebral malformations.[11]

Second trimester prenatal diagnosis[6] was done in 4 of our 13 cases. A deformed spine and incompletely formed ribs were demonstrated in all cases. Prenatal diagnosis is thus feasible and important to detect the more aggressive and lethal form of JLS, *i.e.*, STD.[1]

Genetic counseling is likewise fundamental. In the recessive form of SCD, the risk to siblings of an affected child is 25%. In the dominant form, this risk is negligible but rises to 50% for the offspring of a long survivor.[1,12]

The survival rate seen in our 2- to 6-year follow-up was 100%. This applies to a series of patients affected by both the dominant and the recessive form of SCD. The recessive form of the disease resulted from a number of patients being born to consanguineous parents (Table 1).

The outcome of conservative treatment was noteworthy in all cases, especially considering the dramatic decrease in the incidence of respiratory infections. Both CPT and hospitalization had to be repeated at regular intervals. Noncompliance was noted in some cases, especially after the reestablishment of the weight curves observed with improved general conditions.

Prolonged survival thus leads to issues of chest wall or spinal deformity management. Chest wall reconstruction has been performed by using either polypropylene mesh[13] or, in our case, autologous muscle flap[7] to improve chest wall compliance. The results have been so far favorable. Spinal fusion in our patient adds to the few other relative descriptions[2,8,14] and eventually succeeded in stabilizing a progressive scoliotic deformity with pelvic obliquity.

■ Conclusion

Based on the above findings we feel that assessment and treatment of SCD should be based on the following guidelines:

- Early prenatal ultrasound diagnosis of SCD is feasible and genetic counseling thus plays an important role.
- In view of the concomitant nonskeletal anomalies, a thorough clinical and radiologic screening of every patient with these skeletal defects is essential.
- Early and repeated CPT forms the mainstay of treatment.
- Prolongation of life span with CPT opens up new possibilities for surgical intervention for reconstruction or deformity correction.
- Surgical intervention for correcting costal defects and spinal surgery with instrumentation seem to be beneficial in these cases.

■ Key Points

- Spondylocostal dysostosis is a form of hereditary scoliosis amenable to prenatal ultrasound diagnosis.
- A mainstay of treatment is conservative with chest physiotherapy plus prevention and treatment of respiratory tract infection.
- Surgery is reserved to selected cases of untreatable respiratory deterioration or scoliosis progression.

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