Tethered Cord in a Patient with Multiple Vertebral Segmentation Defects
A Case Report

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Abstract
Short trunk dwarfism with multiple vertebral segmentation defects (MVSD) represents a heterogeneous group of disorders characterized by the presence of multiple vertebral and rib abnormalities. A two and one-half year-old female with the spondylothoracic dysostosis form of MVSD is presented. In addition to skeletal anomalies, a lumbar hemangioma, bilateral foot deformities, distal leg atrophy and weakness, and areflexia at the ankles were present. An underlying neuropathic process was suspected. Results of urodynamic studies were suggestive of a neurogenic bladder. Magnetic resonance imaging of the spine demonstrated a tethered spinal cord. Although various brain and spinal cord anomalies have been described in MVSD, this is the first reported case, to our knowledge, of a tethered spinal cord in a patient with MVSD. We recommend that the management of patients with MVSD include comprehensive neurological evaluation and monitoring with appropriate electrodiagnostic, urodynamic, and neuroimaging studies.

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hort trunk dwarfism with multiple vertebral segmentation defects (MVSD) represents a heterogeneous group of disorders characterized by short stature and multiple vertebral and rib anomalies. Variable clinical findings include a shortened neck with low posterior hairline, kyphoscoliosis, pectus deformities, and respiratory difficulties.

The classification of patients with this condition remains controversial. Several classification schemes have been proposed.1-3 Recently, Mortier and colleagues4 have delineated three forms on the basis of radiographic and clinical findings and on patterns of inheritance. The first form, also known as Jarcho-Levin Syndrome, has an autosomal recessive pattern of inheritance and is characterized by a symmetrical crab-like configuration of the ribs, segmentation defects throughout all vertebral levels, absence of other major congenital anomalies, and early death due to respiratory problems. This form has an increased frequency in the Puerto Rican population.

The second form, called spondylothoracic dysostosis, also has an autosomal recessive pattern of inheritance and is characterized radiographically by fusion and/or absence of ribs and multiple segmentation defects involving all vertebral levels. Variability in terms of severity ranges from death in infancy as a result of respiratory failure to minimal symptoms with survival into adulthood. Associated anomalies and respiratory difficulties are absent.

The third form, called spondylocostal dysostosis, is characterized by an autosomal dominant pattern of inheritance and normal life span. Spine and rib abnormalities are similar to but usually milder than those present in spondylothoracic dysostosis. Affected individuals typically present after infancy with problems associated with kyphoscoliosis, low back pain or decreased spinal mobility. Associated anomalies and respiratory difficulties are absent.
Sporadic cases are often difficult to classify due to the overlapping of clinical and radiographic findings between the latter two forms. Associated abnormalities are more commonly present in sporadic cases.

Associated non-skeletal anomalies tend to correspond to the level of the vertebral defects and include bilobed bladder, hydronephrosis, hernia, anal and urethral atresia, and talipes equinovarus. Additionally, neurological abnormalities such as cerebral polygyria and neural tube defects have been described. To our knowledge, the present case is the first report of a child with multiple vertebral segmentation defects and a tethered cord.

Case Report

The patient is a two and one-half year-old female with MVSD, born to a 23-year-old G3P2 Puerto Rican female and a 17-year-old African-American male. No parental consanguinity was present and maternal and paternal family histories were unremarkable.

The pregnancy was reportedly uneventful until approximately 4.5 to 5 months of gestation when spinal anomalies were noted on the prenatal ultrasound. The pregnancy was continued to term. A Cesarean section was performed secondary to fetal bradycardia. Length at birth was 34.3 cm (significantly below the 5th percentile) and birth weight was 2550 gm (approximately the 10th percentile). The infant cried immediately and did not require ventilatory assistance. Spine and limb anomalies were noted at birth. The infant developed hyperbilirubinemia which was treated with phototherapy. The infant was discharged home at two weeks of age.

The patient was referred to the Growth Center of the Hospital for Joint Diseases at three months of age. Physical examination demonstrated a head circumference of 41 cm (40th percentile) and birth weight was 2550 gm (approximately the 10th percentile). The infant cried immediately and did not require ventilatory assistance. Spine and limb anomalies were noted at birth. The infant developed hyperbilirubinemia which was treated with phototherapy. The infant was discharged home at two weeks of age.

The patient was referred to the Growth Center of the Hospital for Joint Diseases at three months of age. Physical examination demonstrated a head circumference of 41 cm (40th percentile), and length of 51.5 cm (significantly less than the 3rd percentile). She had a round face with deep nasal bridge, long philtrum, anteverted nostrils, thin upper lip, short neck, short chest, protuberant abdomen, diastasis recti, normal female genitalia, and no organomegaly. Her spine was short with a hemangiomata in the lumbar region and a gibbus at the thoracolumbar junction.

Abnormalities noted on spine and rib radiographs (Fig. 1) included:
1. Shortened thoracic spine with mild thoracic dextroscoliosis and lumbar levoscoliosis,
2. Multiple fused thoracic vertebrae with at least one absent or markedly hypoplastic upper thoracic vertebra,
3. Anterior hemivertebrae at L1 and L2 that were displaced posteriorly resulting in mild to moderate central stenosis and focal kyphosis at the thoracolumbar junction,
4. Anterior hemivertebra at S1 with posterior subluxation resulting in mild narrowing of central canal and loss of lumbar lordosis,
5. Spinal dysraphism from T10 to S2 with widened interpediculate distance and lack of fusion of posterior elements, and
6. Nine ribs on the left and 10 ribs on the right with hypoplasia of the fourth rib and posterior fusion of fifth, sixth, and seventh costal arches.

Radiographs of the lower extremities demonstrated bilateral coxa valga, a right vertical talus and a left clubfoot.

Motor examination of the upper extremities was normal. Examination of the lower extremities revealed distal atrophy, left equinovarus deformity with foot deformity, and a right calcaneovalgus deformity. Limitations in passive hip internal rotation and adduction, and bilateral hip and knee flexion contractures were noted. Voluntary flexion and extension of the knees were limited with minimal movement of feet and toes. Deep tendon reflexes were 2+ in upper extremities, 1+ to 2+ at knees, and absent at both ankles. Plantar responses were flexor bilaterally.

Electromyography (EMG) and nerve conduction velocity studies were remarkable for absent H reflexes bilaterally. Cortical SSEPs following stimulation of the posterior tibial and peroneal nerves showed delayed latencies. These findings were suggestive of L5-S1 segmental pathway dysfunction.

Magnetic resonance imaging (MRI) performed at 13 months of age demonstrated kyphoscoliosis of the thoracolumbar spine, spondylolisthesis at L5-S1, and a tethered cord at L5. No other abnormalities of the spinal cord were noted.

Renal sonogram revealed normal sized kidneys without evidence of gross abnormality. Results of urodynamic studies were suggestive of a neurogenic bladder.

In view of the clinical, electrophysiologic, urologic, and neuroimaging findings, the patient underwent detethering of the cord at 20 months of age. A fibrous band attached to the dorsum of the spinal cord at approximately the S1-S2 segment level and a tight filum were released.

Discussion

The multiple vertebral and rib malformations in our patient are consistent with the diagnosis of MVSD. Since our patient represents a sporadic case of MVSD, definitive classification is difficult. We feel that the clinical and radiographic abnormalities in our patient fit best a diagnosis of spondylothoracic dysostosis.

In addition to skeletal deformities, a lumbar hemangiomata, bilateral foot deformities, distal leg atrophy and weakness, and areflexia at the ankles were present in this patient. These associated abnormalities corresponded to the level of her vertebral abnormalities. An underlying neuropathic process was suspected. The results of
Urodynamic studies were suggestive of a neurogenic bladder. Magnetic resonance imaging of the spine revealed the presence of a tethered cord.

Multiple structural abnormalities of the spinal cord and brain have been described in patients with MVSD including cerebral polygyria, diastematomyelia, Arnold-Chiari malformation with thoracolumbar meningomyelocele, thoracolumbar rachischisis and hydrocephalus due to aqueductal stenosis, spina bifida cystica in the thoracolumbar region, and diastematomyelia. A patient with MVSD and progressive thoracolumbar kyphosis with concomitant neurological impairment of the lower extremities was reported. Anteroposterior spinal fusion and decompression were required. To our knowledge, our patient represents the first documented case of a tethered cord associated with MVSD.

At birth, the spinal cord terminates at the L3 level. Over the first six months of life, the vertebral elements grow more than the neural elements, causing the spinal cord termination to migrate up to the normal adult level of L1-L2. The conus medullaris (spinal cord termination) is normally anchored to the end of the dural sac by the filum terminale, which is a fibrous band originating from the distal termination of the pia mater. The slack in the filum permits spinal flexion without stretching the distal cord. In pathological cases, the filum is abnormally thickened. The lack of elasticity of the filum causes the spinal cord to stretch during growth. Distal spinal cord dysfunction may occur as the child’s trunk lengthens or following a growth spurt.

Patients with a tethered cord may be divided into two groups:

1. An asymptomatic group investigated because of a high index of suspicion (i.e., hemangioma, tuft of hair, dimple above gluteal cleft) or incidentally found to have abnormal neuroimaging studies; and
2. A symptomatic group presenting with leg or back pain, urinary incontinence, neurologic signs, and/or orthopaedic deformities.

The back or leg pain in these patients is persistent and exacerbated by exercise or spinal flexion. Orthopaedic manifestations include scoliosis, various foot deformities, leg length discrepancies, and chronic hip dislocation. Bladder and bowel dysfunction may present as failure to toilet train, loss of control, or urinary/fecal urgency or incontinence. Neurologic manifestations include lower extremity weakness, gait abnormalities, and loss of proprioception in the legs.

Historically, patients with severe MVSD have had a high infant mortality rate due to early respiratory compromise. Advances in respiratory management have improved the prognosis for survival. Patients with this syndrome surviving beyond five years of age are predisposed to progressive scoliosis and neurologic dysfunction.

As previously noted, multiple brain and spinal cord anomalies have been described in patients with MVSD. Based on the findings in our patient, we suggest that tethered cord be added to the list of spinal cord abnormalities. The presence of vertebral segmentation defects in any patient should raise a high index of suspicion for a tethered spinal cord, which may be a cause of progressive neurological dysfunction. Management of patients with MVSD should include comprehensive neurological evaluation and monitoring with appropriate electrodiagnostic, urodynamic, and neuroimaging studies.

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References