Case Report

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Spinal and pelvic corrections in a patient with spondylocostal dysplasia syndrome and hemimyelomeningocele

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ABSTRACT

Congenital malformation complex of the spine and the spinal cord can be a syndromic entity rather than a symptom complex. The spinal cord lesion is usually bilaterally symmetrical, but, there are occasional cases with one or more hemivertebrae, often associated with a central bony spur splitting the cord (diastematomyelia), in which one leg is virtually normal while the other is severely paralysed. Hemimyelomeningocele over the lumbar area may be associated with extensive spine malsegmentation compatible with the diagnosis of spondylocostal dysplasia syndrome. In this report, we present a 3-year-old girl who underwent neurological evaluation and spinal imaging studies for extensive spine malsegmentation compatible with spondylocostal dysostosis syndrome associated with hemimyelomeningocele. She had a series of corrective orthopaedic interventions to reconstruct her pelvic girdle and spine deformities, with a satisfactory outcome.

Key words: Computed tomography scan, hemimeyelomeningocele, magnetic resonance imaging, spondylocostal dysplasia syndrome, spondylopelvic malformation

INTRODUCTION

Disruptions in somitogenesis cause vertebral malformations, including failure of formation and segmentation such as hemivertebrae and wedge vertebrae, block vertebrae or butterfly vertebrae.[1]

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Congenital vertebral defects that result from disruption of the induction and formation of the axial skeleton include spondylocostal dysostosis (SCD), Jarcho-Levin syndrome (JLS), Klippel-Feil anomaly and a wide range of syndromic malformation complex such as VATER, VACTERL and MURCS association.[1-3]

Jarcho-Levin syndrome is a congenital dysostosis that affects the bones of the spine and the ribs. The main features are short stature with a very short thorax, rib cage abnormalities and multiple vertebral anomalies. It is an autosomal recessive disorder, and spina bifida is present in 25% of patients. Morbidity and mortality among patients with JLS has been attributed to congenital thoracic cage abnormalities.[2,3]

Karnes *et al.*,[4] reviewed the differences between spondylocostal dysplasia and JLS. Nevertheless, some cases reported as JLS probably have a form of spondylocostal dysplasia. It has been suggested that the pathogenetic aspect in correlation with the radiographic pattern of presentation are the main hallmarks for classification of SCD.[5] Previous studies investigated the reason of death in patients with spondylothoracic dysplasia and spondylocostal dysplasia (SCD). A congenital thoracic cage abnormality with concomitant progressive thoracic cage restrictions and development of thoracic insufficiency syndrome was the main reason thought to be responsible for the high fatality rate in these patients.[6]

Neural tube defects are considered to result from the failure of normal neural tube closure between the 3rd and 4th week of embryonic development .In general, they vary in severity, the mildest form being spina bifida occulta, in which osseous fusion of one or more vertebral arches is lacking, without the involvement of the underlying meninges or neural tissue. This lesion is covered by skin, therefore, rendering the underlying neurologic involvement occult or hidden.[7] The association of segmental costovertebral malformations and neural tube

defects has been reported, and it has been proposed that this association is not coincidental.[8-10] Incomplete ossification of the cervico-thoracic pedicles associated with extensive malsegmentation is a major reason for the development of progressive kyphosis in patients with diastrophic dysplasia, chondrodysplasia punctata, Larsen syndrome and camptomelic dysplasia.[11]

Advanced scanning techniques can provide additional information which facilitates orthopaedic interventions. The prognosis for deterioration of the severe spinal deformity in this patient was anticipated due to the insufficient spinal growth of the concavity. The type and the site of the vertebral anomalies and the degree of growth imbalance were extensive.

CASE REPORT

A 3-year-old girl was referred to the orthopaedic department due to progressive lumbosacral scoliosis associated with a significant tilt of the lumbosacral/sacroiliac complex and neurogenic right side club foot. She was born full term. At birth, her birth weight, length and ofc were around the 10th percentile. Short trunk dwarfism associated with congenital lumbar scoliosis, and myelomeningocele were the prominent clinical features. Anteroposterior (AP) radiograph showed extensive malsegmenation of the entire spine associated with congenital scoliosis and dislocation of the right hip [Figure 1]. AP spine radiograph revealed extensive malsegmentation of the entire spine ranging between butterfly vertebrae to hemivertebrae associated with neurocentral synchondrosis of the entire cervical spine [Figure 1].

Hemimyelomeningocele (HMM) was diagnosed, and a lumbar lipoma was recognized via magnetic resonance imaging (MRI) intradural lipoma. Sagittal T1-weighted and sagittal T2-weighted fat-saturated showed large intradural lipoma (a split cord malformation secondary to intradural lipoma, and the development of a tethered cord) (*arrows*), which is hyperintense on T1-weighted image and hypointense on T2-weighted fat-saturated image. Lipoma was attached to *conus medullaris*, which is low-lying [Figure 2]. Three-dimensional coronal computed tomography (CT) scan of the lumbo-sacral showed extensive defects of formation of the entire area (hemivertebrae), spondylopelvic dissociation and dislocation of the right hip joint and lateral displacement of the malsegmented sacrum [Figure 3].

The mother was a 25-year-old gravida 1 abortus 0, married to a –29-year-old unrelated man. Family history



Figure 1: AP spine radiograph revealed extensive malsegmentation of the entire spine ranging between butterfly vertebrae to hemivertebrae associated with neurocentral synchondrosis of the entire cervical spine

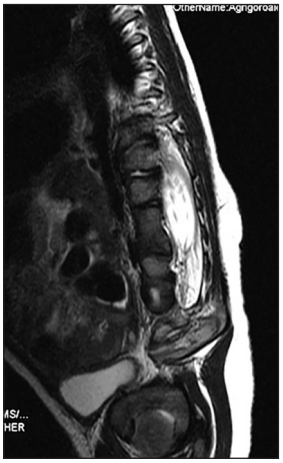


Figure 2: Hemimyelomeningocele was diagnosed, and a lumbar lipoma was recognized via MRI Intradural lipoma. Sagittal T1-weighted and sagittal T2-weighted fat-saturated showed large intradural lipoma (a split cord malformation secondary to intradural lipoma, and the development of a tethered cord) (*arrows*), which is hyperintense on T1-weighted image and hypointense on T2-weighted fat-saturated image. Lipoma was attached to *conus medullaris*, which is low-lying

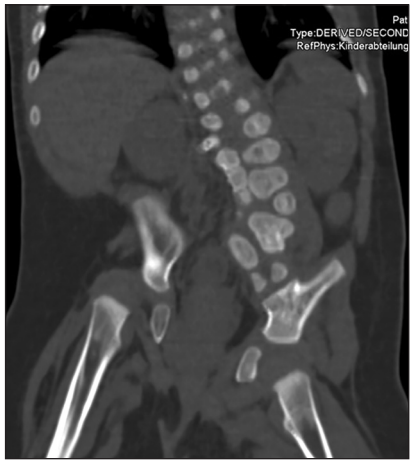


Figure 3: Three-dimensional coronal computed tomography scan of the lumbo-sacral showed extensive defects of formation of the entire area (hemivertebrae), spondylopelvic dissociation and dislocation of the right hip joint and lateral displacement of the malsegmented sacrum

revealed the presence of vertebral malsegmenation in the father and her cousin (paternal side). Investigations included creatine kinase plasma levels, metabolic screening of plasma and urine for disturbances in the metabolism of amino acids, bile acids, phytanic acid and oligosaccharides, all gave normal results. There were normal serum levels of luteinizing hormone, prolactin and estradiol. Blood sugar, uric acid levels, serum calcium, phosphorous and parathyroid hormone levels were within normal limits. Chromosomal analysis in both the patient and the parents revealed normal karyogram. Echo-Cardio-Doppler was normal. Her vision, hearing and intelligence were normal. Neurological examination was normal. We were unable to perform genetic study on this family due to logistical reasons.

Examination at the age of 3 years showed a girl with short trunk dwarfism (−4 SD). No peculiar craniofacial dysmorphic features were encountered. Her hands were normal, but nevertheless, short trunk dwarfism was the most prominent feature. Significant spinal malformations complex were noted. Hearing, vision and intelligence were normal. Options for surgical treatment in the presence of lumbosacral malformations were limited, as the malsegmented sacrum did not offer possibilities of instrumentation.

Phenotypic characterization followed by a series of corrective orthopaedic interventions to reconstruct her pelvic girdle and spine deformities was performed. She had correction of her spine deformity by removal of unilateral lumbar double hemivertebra and lumbosacral fixation.

The objective of treatment was to improve walking ability and trunk balance. An attempt was made to balance her spinal growth by removing abnormal spinal growth at the convexity. This patient presented with severe failure of segmentation and formation of the entire spine compatible with spondylocostal dysplasia associated with hemimeylomeningocele.

Treatment

For the severe neurogenic right side clubfoot, a complete peritalar release according to McKay and Simons was performed at the age of 1 year. The medial, dorsal and lateral aspects of the foot were released from ankle joint and subtalar joint by a Cincinnati approach. After correction of the subtalar joint and the chopard joint, line fixations with 3-K wires and casting for 6 weeks were realized, followed by treatment with orthosis.

For the dislocation of the right hip, open reduction of the right hip was performed at the age of 2 years using an inguinal approach, a tenotomy of the psoas tendon and a percutaneous release of the adductor muscle were included. T-shaped opening of the capsule with removal of all soft tissues inside the acetabulum was done. A lateral approach of the proximal femur with shortening and varisation osteotomy was performed to reduce the pressure of the femoral head inside the acetabulum. Pemberton acetabuloplasty was added to improve the coverage of the femoral head. An apica cast was applied for 6 weeks. The hardware (plates and K-wires) were removed 8th months later [Figure 4].

For the spine deformity (for the left side convexity of the lumbar spine and because of the extensive malsegmentation), de-tethering was performed in the 1st year of life. In 21-08-2012, resection of the hemivertebrae of L3-4-5. Posterior fusion of L2-S1 was accomplished, and a spica cast was applied [Figure 5]. The image of a child after surgery at age of 4 years showed leg length discrepancy and hypotrophy of the right lower limb, recurrence of right equinus deformity (varus of the hind foot and supination of the forefoot, respectively) [Figure 6]. At last follow-up, the girl can walk independently using a shoelift of 7 cm to compensate her leg length discrepancy. The shortening is partly (4 cm) caused by the pelvic obliquity due to the spinal malformation. The hip joint on the right is

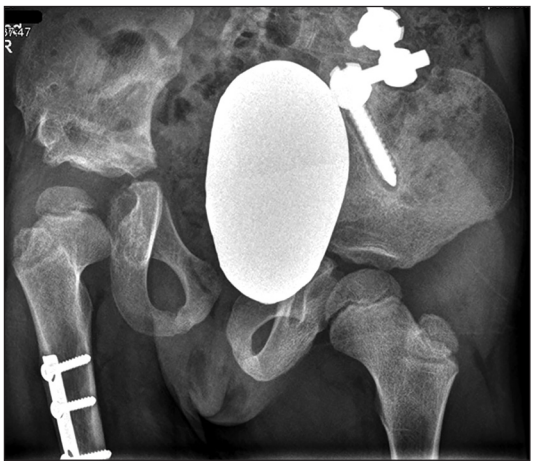


Figure 4: For the dislocation of the right hip, in 3.11.2010, open reduction of the right hip was performed using an inguinal approach, a tenotomie of the tendon of the psoas muscle and a percutaneous release of the adductor muscle was included. A T-shape opening of the capsule was performed, after that removal of all the soft tissues inside the acetabulum was done. A lateral approach of the proximal femur shortening varisation was performed to minimize the pressure of the femoral head inside the acetabulum was done. The osteotomy was fixed with an AO miniplate. The latter procedure was followed by reposition of the femoral head inside the acetabulum, simultaneously with the closure of the hip joint. Acetabuloplasty according to Pemberton was applied to ameliorate the coverage of the femoral head. K-wires were used to affirm fixation, and spica cast for 6 weeks was used thereafter. The hardware (plates and K-wires) were removed in July 2013

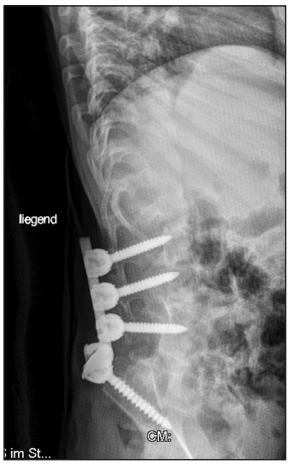


Figure 5: For the spine deformity (for the left side convexity of the lumbar spine and because of the extensive malsegmenation), de-tethering was performed in the 1st year of life. In 21.8.2012, resection of the hemivertebrae of L3-4-5. Posterior fusion of L2-S1 was made, and spica-cast was applied accordingly



Figure 6: The image of a child after surgery at age of 4 years showed leg length discrepancy and hypotrophy of the right lower limb, recurrence of right equinus deformity (varus of the hind foot and supination of the forefoot, respectively)

still well-reduced. There is a recurrence of equinovarus deformity of the right foot, which needs additional surgical correction. The foot is stiff, active movements of the toes are possible.

DISCUSSION

The development of the spinal cord is closely associated with that of the vertebral column. It is, therefore, not surprising that neural and vertebral malformations often coexists. Spine malformations are frequently associated with congenital anomalies of other systems, especially those formed from mesenchyma. These anomalies are either symptomatic or asymptomatic. The genito-urinary system may be affected in up to 25% of patients. A unilateral kidney is the most common associated anomaly. Duplication of the kidney or ureteric obstruction also do occur. In order to understand the natural history of congenital spine malformations and the great variety in prognosis, it is necessary to correlate the principles of normal growth of the spine with the pathologic anatomy of the various types of congenital vertebral anomalies.[7,9]

Kusumi and Turnpenny[1] described the different syndromes that occur due to failure of formation of the spinal column. They emphasised the significant role of the radiographic interpretation in classifying the different forms of SCD, but three-dimensional scanning studies were not included. Turnpenny *et al*.[11] reported the clinical, radiographical and molecular findings in 10 families with autosomal recessive SCD. Radiologically, there was abnormal segmentation throughout the entire vertebral column with smooth outlines of the vertebral bodies in childhood. Further characterisation of the spinal pathology by means of other scanning modalities was not applied. For the first time, Al Kaissi *et al*.,[12] provided a detailed craniocervical analysis via 3DCT scan in SCD patients.

Mutations have been identified in four genes involved in the Notch signalling pathway: *DLL3, MESP2, LFNG,* and *HES7*. Alterations in these genes produce the autosomal recessive variety of SCD, but have been excluded in the dominant SCD families. There are some reports of heterozygous mutations in *GDF3* and *GDF6* as causing dominant SCD. MESP2-associated SCD (SCD2) showed a severe generalized vertebral disorder, with some patients having ribs that radiate out in what has been described as "crab chest" and some have referred to as "Jarcho-Levin" syndrome in which the ribs are more compressed but may have less severe modelling abnormality than the SCD1 patients. Mutation in LFNG-associated SCD type 3 also showed a severe generalised vertebral involvement, often with a significant reduction in spine height. Mutations in *HES7*-associated SCD type 4 have been reported in some patients, and their radiographs are similar to SCD 3.[13] Furthermore, mutations in DLL3-associated spondylocostal dysostosis type 1 (SCD1) is a form with the so-called "pebble beach" sign, that is vertebral bodies are irregular in size but have a smoothly rounded shape. The thorax tends to be symmetric and, despite the significant vertebral abnormalities, there is only mild scoliosis. Mutations in *MESP2*-associated spondylocostal dysplasia type 2 (SCD2) showed a severe generalized vertebral disorder, with some patients having ribs that radiate out in what has been described as the "crab chest" and some have referred to as " JLS".[13] Rodriguez *et al*.[14] reported the association

of SCD with perinatal death and meningomyelocele. Etus *et al*.[8] described the association of spondylocostal dysplasia and type I split cord malformation None of the above-mentioned reports described the association of SCD and hemimyelomeningocele with subsequent maldevelopment of spondylopelvic malformation.

In previous reports, the term JLS and SCD were used interchangeably. The association of segmental costo-vertebral malformations and neural tube defects has been reported, and it has been proposed that this association is not coincidental. Reyes *et al*.[15] reported a case of Jarcho-Levin syndrome with diastematomyelia of the thoracolumbar spinal cord and suggested neural tube defects as a component of JLS. Duru *et al*.[16] reported two patients with JLS and neural tube defects, one of whom with lumbosacral lipomyelomeningocele and the other with thoracolumbar myelomeningocele.

Hemimyelomeningocele refers to a unique form of spinal cord dysraphism in which the myelocele or myelomeningocele is found in one hemicord of a diastemmatomylia. Hemicords usually lie in their own dural tube and are separated in the midline by a bony septum.[17] The myelomeningocele is known to be a defect of primary neurulation with the lack of midline closure of the neural placode and contiguous structures. It takes place at the 26-28 days of gestation.[18] On the other hand, diatsematomyelia is basically known to be a defect of secondary neurulation or postneurulation.[19] HMM is observed in about 10% of unselected patients with myelomeningocele.[20] The neurological deficit in children with HMM is less severe than compared to that seen in myelomeningocele patients. It has been reported that children with HMM have neurological deficits limited to the exposed hemicord and have normal function on the side of the normal hemicord.[17-20]

The split-cord malformation or (diastemmatomyelia) represents a sagittal cleft of the spinal cord into two hemicords that can be enclosed in single or separate dural sheaths and separated by a fibrous or osteocartilaginous septum, respectively. The condition is usually accompanied by a low tethered conus and thick filum terminale.[21]

IN SUMMARY

The reason for presenting this case is four-fold. First, no previous reports described the association of hemimyelomeningocele and spondylopelvic deformity in connection with SCD. Second, by using 3DCT, we were able to visualize the severe defects of formation (hemivertebrae) along the lumbar area and the associated lumbo-sacral pathology and the right hip dislocation. Third, it was extremely difficult to approach optimum surgical results due to the severity of the spinal deformity and the pelvic malformation. Fourth, the father and cousin were also affected by SCD raising the possibility of autosomal dominant pattern of inheritance.

Proper documentation of children with multiple malformations along with the help of CT scans provides excellent delineation of osseous deformity patterns in children with syndromic association. Three-dimensional studies and sagittal and coronal reconstructions provide the orthopaedic surgeon with a detailed understanding of the spinal osseous deformity and the development of the subsequent concomitant atrophy of the spinal cord. These investigations have important implications in understanding the mechanism of the patho-anatomy, judging potential corrective measures, evaluating deformity and planning surgical procedures. We conclude that the use of the advanced scanning techniques, in combination with plain radiographs, will provide the orthopaedic surgeons with an appropriate understanding of the pathologic and surgical anatomy present in most patients with vertebral malformations. The relationship of the spine to the sacrum and the iliem should be carefully assessed in order to correct the spondylopelvic malalignment. Our observations may contribute to support early diagnosis and treatment. Failure to accurately detect these abnormalities can lead to serious orthopaedic/neurologic complications.

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