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Case Report

Segmental Spinal Dysgenesis: A Case Report

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Abstract

Segmental spinal dysgenesis (SSD) is a rare and complex congenital anomaly of the lumbar or thoracolumbar spine which is associated with congenital kyphosis or kyphoscoliosis and focal spinal cord malformations. In this study, we describe a 5 day old newborn with congenital paraparesis, talipes equinovarus and complex kyphoscoliosis diagnosed via fetal ultrasound. The diagnosis of SSD was confirmed by post-natal CT scan and MRI which showed a complex malformation at the thoracolumbar junction along with spinal cord hypoplasia. The baby underwent rehabilitation alongside a thoracic-lumbar orthosis to prevent progressive kyphotic deformities. Surgical spine correction and fixation was planned at the 2 year old mark, once an adequate bone maturity was achieved. The present study highlights the peculiar clinical picture of this extremely rare syndrome, providing new insights about the clinical diagnosis and management.

INTRODUCTION

Segmental spinal dysgenesis (SSD) is a rare and complex congenital spinal anomaly characterized by localized agenesis or dysgenesis of the lumbar or thoracolumbar spine, congenital kyphosis and kyphoscoliosis, and focal abnormalities of the underlying spinal cord and nerve roots. The malformation is usually segmental with normal vertebrae above and below the abnormal segment. The affected spinal cord can be both severely hypoplastic or incompletely developed and interrupted at the level of malformation [1-3]. The clinical picture of these young patients consists in motor impairment, from mild deficit to paraplegia, and depends on the severity of malformation of the spinal cord, which can range from moderate hypoplasia to complete absence, and on the degree of residual function. Deformities of the lower limbs and neurogenic bladder can be also associated, the latter leading to increased risk of urinary incontinence and complications such as uni- or bilateral vesico-ureteral reflux and urinary infections [3-6].

CASE REPORT

In June 2022, a 5 day old male newborn was admitted to our Neonatal Intensive Care Unit (NICU), with a clinical picture of talipes equinovarus and complex kyphoscoliosis. Prenatal diagnosis was made at the 34th week of gestational age via fetal ultrasound. The mother was a young Ukrainian woman with no medical history, except for SARS-CoV2 infection in the II trimester of pregnancy. She did not take folic acid during the pregnancy due to the lack of prescription.

On admission, the newborn presented pes equinovarus, overt kyphoscoliosis, severe distal arthrogryposis, and paraparesis. No spontaneous movements of the lower extremities were observed. The baby had inconsistent and limited flexion movements on the pelvis, most likely due to myoclonic jerks, absent deep tendon reflexes at the lower limbs, and spontaneous micturition and defecation, suggesting a preserved sphincteric function. Movements of the upper limbs and upper trunk were preserved.

The diagnostic workup included blood tests, cardiac and cranial ultrasounds, audiological and ophthalmic evaluations, all of which resulted unremarkable. Brain and spinal cord MRI performed on the 13th day after birth showed a complex malformation at the thoracolumbar junction, with dysmorphic and apparently fused vertebral metameres, which appeared rotated and laterally displaced. The dural sac and spinal cord were not recognizable at the level of the malformation, being discernible only from the skull base up to T5 and caudally to the

malformation (Figure 1). That malformation was explained as a suspected segmental spinal dysgenesis. No clear syringomyelia was detected. The 3D-CT scan confirmed the aplasia of T9 to T12 metameres (Figure 2). On this radiological basis, the picture of congenital severe paraparesis was consistent with the malformation. To investigate the residual function of the spinal cord, four-limbs somatosensory evoked potential (SSEP) were performed which confirmed the absence of nervous conduction across the segments involved by the malformation. Given the absence of a vital spinal cord at malformation level, no decompression spine surgery was recommended, and the child started the application of a thoracic-lumbar orthosis to avoid progressive kyphotic deformity. Late surgery of spine fixation was planned at around 2 years-old, once the adequate spine growth and bone are achieved.

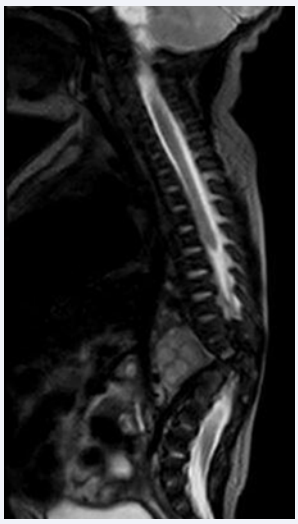

Figure 1 Sagittal T2-weighted MR image of brain and spinal cord performed at day 13 after birth, which shows a complex malformation at the thoracolumbar junction, with dysmorphic and apparently fused vertebral metameres and not clearly recognizable dural sac and spinal cord, which appeared to be discernible only from the skull base down to T5 and caudally to the malformation.

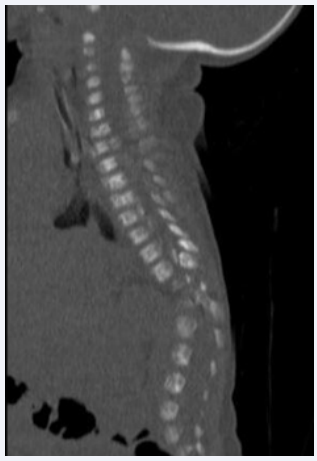

Figure 2 Sagittal section CT-Scan of the vertebral column shows the absence of vertebrae from T9 to T12 and the absence of discernible spinal canal cord from T8 to L1

The genetic work-up did not reveal any specific syndrome but the exposure to teratogenic agents during pregnancy was allegedly suspected.

Regarding the lower limb arthrogryposis, the baby was initially treated with "Ponseti method," using manipulation and plaster casts to maintain the feet in the correct position.

After a two-months hospitalization, the baby was discharged from the NICU and the family was recommended performing neurosurgical, orthopedic, neurologic and rehabilitative follow-up.

At the 9 month reevaluation, the patient was overall in good condition. Since the discharge, he had presented only one episode of urinary tract infection, treated with antibiotic therapy. He had worn the thoracolumbar corset and foot braces which had provided the anticipated clinical benefit. A follow-up evaluation with spine imaging has been scheduled to reassess his condition. Regarding the neuromotor and neurodevelopmental aspects, the child has continued physiotherapy training twice a week; at reevaluation upper limb motility was preserved while lower limb motor function was still poor. Specifically, he was able to perform hip and knee flexion movements bilaterally. Segmental ankle and toe movements could be elicited only after stimulation. If facilitated, he was able to rotate from supine to prone and to sit with support.

DISCUSSION

The present paper adds a new case of complex SSD diagnosed in a newborn. In 1988, Scott et al. [1], described and recognized the first case of SSD as an autonomous entity. According to their definition of SSD, the following diagnostic criteria should be fulfilled: segmental agenesis or dysgenesis of the lumbar or thoracolumbar spine, segmental abnormality of the underlying spinal cord, congenital paraplegia or paraparesis and congenital lower limb deformities. Subsequently, other authors and studies helped to characterize the clinical and neuroradiologic features of this syndrome [2,3].

Neuroradiologic findings include localized deformity of the spine associated with abnormal development of the underlying spinal cord and nerve roots [1]. This malformation is mostly observed at the thoracolumbar, lumbar, or lumbosacral zone; although, cases of cervicothoracic or multisegmental spinal cord involvement have also been reported [7,8]. The level of conus medullaris is often lower than normal range and a large spectrum of congenital kyphosis- from mild to severe- may be associated [3]. MRI is the first choice for neuroradiological investigation while CT is complementary in further detailing the bone anatomy [2,9]. In our case, in line with current literature, the neuro-radiological picture from the MRI demonstrated a complex malformation. This consisted in dysmorphic and apparently fused vertebral metameres at the thoracolumbar junction, with barely not recognizable dural sac and spinal cord: the latter being discernible only from the skull base up to T5 and caudally to the malformation. Furthermore, the CT scan (Figure 2) as well as

the subsequent 3D-CT reconstructions of the vertebral column (Figure 3), confirmed the aplasia of T9 to T12 metameres.

The clinical picture of these young patients is already apparent at birth and consists in motor impairment, ranging from mild deficit to paraplegia. The severity of neurologic impairment depends on the severity of malformation of the spinal cord which can range from moderate hypoplasia to complete absence, thus depending on the degree of residual function. Deep tendon reflexes are often reduced or absent, wherein various deformities of the lower limbs can be associated, and neurogenic bladder is also often present. The latter predisposes the patient to urinary incontinence and increases the risk of complications such as unilateral or bilateral vescico-ureteral reflux and urinary infections [3–6]. SSD has been described in association with other spinal abnormalities that fall within the spectrum of closed spinal dysraphisms, such as diastematomyelia or split cord malformation (SCM), terminal myelocystocele, filar lipomas, and thickened filum terminale as well as with kidney malformations like horseshoe kidney and renal ectopia [1,10]. Conversely to literature data, in the case of the aforementioned patient none of these expected spinal abnormalities were found.

Considering the etiopathogenesis, literature data suggest that SSD may be teratogen-induced or genetically imprinted [2]. An error occurring during gastrulation is the most widely accepted hypothesis [11]. Specifically, from day 14 to 15 of the embryogenesis, a complex mechanism involving an abnormal cell migration between the ectoderm and the interposed mesoderm, or a genetically induced alteration in elimination apoptosis, can lead to several varieties of dysraphic states. This can include the full blown split notochord syndrome, diastematomyelia, dermal sinus tracts, caudal agenesis, and SSD [12-15].

The variety of the clinical and neuroradiological picture depends on the severity of the damage and neuroectodermal abnormalities, the localization of the segmental dysgenesis along the spinal cord, and the residual function of the malformed segment [2,3]. In our case, the patient, on admission at the NICU at 5 days old, presented a clinical picture of distal paraparesis, consisting in lack of spontaneous movements of the lower extremities and absent deep tendon reflexes at the lower limbs; at a subsequent follow-up evaluation at 9 month, he showed slight improvement of lower limbs movement: being able to perform hip and knee flexion movements bilaterally, and segmental ankle and toe movements. Upper limb and upper trunk motility was completely preserved. These features are in line with his neuroradiological picture of the RMN and TC scan, describing the absence of vertebrae from T9 to T12 and the absence of discernible spinal canal cord from T8 to L1.

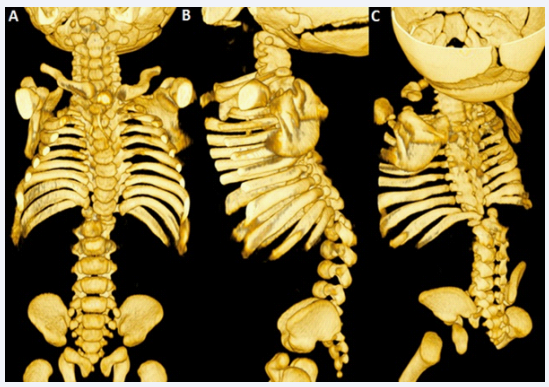

Figure 3

Recently a new SSD classification has been proposed by Chellathurai et al., which subdivides SSD into two types [16]. Type 1 SSD is characterized by congenital segmental absence or malformation of multiple vertebrae, the spinal cord, and its underlying nerve, or by a spinal canal narrowing due to a chordamesoderm positional error. In type 2 SSD, the spinal canal is severely narrowed in all patients, and the spinal cord in the dysgenetic segment is consequently severely compressed, stretched, and thinned-out in segments adjacent to the gibbus apex. It is believed that two different mechanisms underlie the two types: type 1 is characterized by a primary cord hypoplasia while type 2 is a secondary cord hypoplasia due to dysmorphic vertebrae.

As far as surgical intervention is concerned, there is an ongoing debate in the literature about the optimal treatment for these patients and about the correct timing of surgical intervention. Due to the rarity of this complex disease, no guidelines on the type and timing of surgical interventions are available. Even if patients with SSD may be not necessarily paraplegic at presentation, they have a high risk of worsening neurological deficits due to the instability and stenosis of the spine and the potential association with closed spinal dysraphism and subsequent tethered cord syndrome [2,4-6].

Cord decompression has shown uncertain results because the neurologic deficit is often related, at least in part, to cord hypoaplasia rather than to cord compression. Thus, decompressive surgery may be not useful to provide a significant improvement [1].

A different role is played by surgical arthrodesis which addresses the issue of spinal instability and progressive deformity. Arthrodesis is virtually indicated in all SSD cases, and the optimal timing for performing it is a matter of debate. Some authors recommended performing the surgery as soon as possible [4,17], while other authors suggested waiting for the stabilization of the general conditions of the newborn and the maturation of the osseous bed, thus waiting to at least the age of 5-6 months or until 2-3 years [1,5]. This would avoid the risk of failure and reoperation. In the wait for surgical fixation, conservative measures such as bracing are advised by these authors.

The recent reclassification of SSD in two types may help to guide surgical management. In fact, type 1 SSD is characterized by a primary cord hypoplasia and thus decompressive surgery has no role in treating it; in type 2 SSD, however, secondary cord damage due to dysmorphic vertebrae may benefit from surgical

management. The latter may range from simple decompression followed by delayed arthrodesis to a more aggressive strategy, including complete resection of the dysmorphic vertebrae with rib strut grafting and posterior arthrodesis [5,16].

In the case of our patient, we decided not to perform cord decompression because the diagnostic studies showed the absence of a functioning spinal cord at the level of his malformation [1]. We decided to plan a subsequent surgical arthrodesis around the 18 to 24 month of age, once the spine is expected to have reached a sufficient bone maturity. In the meantime, we decided to apply a thoracolumbar corset which prevented progressive kyphotic deformity.

CONCLUSIONS

Our case is in line with literature data and meets all the four inclusion criteria to satisfy the diagnosis of SSD described by Scott et al. [1], since the patient has dysgenesis of the thoracolumbar spine, an underlying segmental abnormality of the correspondent spinal cord, congenital paraparesis, and congenital lower limb deformities.

The present study highlights the peculiar clinical picture of this syndrome. Due to the rarity of this complex disease, no guidelines on the type and timing of surgical interventions are available. More data about the outcomes of these patients needs to be gathered in order to provide an overall view of this rare disease. This report may help provide new insights about the clinical diagnosis of this rare disease and future management.

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