

# Spinal segmental dysgenesis

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## Abstract

Spinal segmental dysgenesis is a rare congenital spinal abnormality seen in neonates and infants, in which a segment of the spine and spinal cord fails to develop normally. The condition is segmental in nature, with vertebrae above and below the malformation. It is commonly associated with various abnormalities that affect the heart and genitourinary and gastrointestinal tracts and skeletal systems. We report two cases of spinal segmental dysgenesis with associated abnormalities.

## Introduction

Spinal segmental dysgenesis (SSD) is a spinal anomaly characterised by localised agenesis or dysgenesis of the lumbar or thoraco-lumbar spine, severe congenital kyphosis or kypho-scoliosis, and focal abnormalities of the underlying spinal cord and nerve roots.<sup>1,2</sup> There have been only a few reported cases of SSD as an autonomous entity. The neuro-radiological presentation is variable depending on the extent and level of the abnormality, the degree of resulting kyphosis and the presence of associated abnormalities. Although computed tomography (CT) has a role in imaging, magnetic resonance imaging (MRI) is the gold standard in the imaging of the spinal cord abnormalities and plays an important role in the planning of surgery.

### Case report 1

A 2-week-old boy was referred from the Paediatric Neurology Department with a history of flaccid paralysis of both lower limbs since birth. The mother described a normal pregnancy with no complications during the pregnancy or the puerperium. Clinically, the child had club feet and was dysmorphic with a scaphoid-shaped back, with clinical suspicion of spina bifida occulta. Spinal X-rays and MRI were requested.

The spinal X-rays (Figs 1a and 1b) demonstrated complete absence of all the lumbar vertebrae and no twelfth thoracic vertebra. In association, only 11 paired ribs were present, and the lower thoracic ribs were flared. The sacrum and pelvis were normally developed. MRI confirmed complete disconnection of the spinal segments above and below the dysgenesis i.e. between T12 to L5 (Fig. 2a). Normal sacrum and sacral nerve roots were demonstrated. An associated abnormality was a horse-shoe kidney (Fig. 2b).

### Case report 2

An 8-week-old boy was referred from the Paediatric Neurosurgery Department with club feet, a sacral dimple and bilateral lower limb paralysis since birth. Spinal X-rays demonstrated complete absence of the lower thoracic, lumbar and sacral spine (Figs 3a and 3b). MRI



Fig. 1a. AP X-ray demonstrates complete absence of all the lumbar vertebrae and twelfth thoracic vertebrae. Eleven paired ribs are present, and the lower thoracic ribs are flared. The sacrum and pelvis are normally developed.

showed that the thoracic vertebrae and spinal cord terminated at T9. There was an associated syrinx from T6 to T9 (Figs 4a and 4b). Below T9 was a linear bony septum which ended abruptly, and an associated dorsal dermal sinus posterior to this. Bilateral malrotated and hydronephrotic kidneys were also present.

## Discussion

Spinal segmental dysgenesis (SSD) is a rare sporadic disorder first described by Scott *et al.* in 1988 (cited in Tortori-Donati *et al.*<sup>1</sup>). This abnormality has also been called medial spinal aplasia, congenital spinal stenosis, or congenital kyphosis and subluxation of the thoraco-lumbar spine owing to vertebral aplasia. The diagnostic criteria include lumbar or thoraco-lumbar spinal dysgenesis or agenesis causing kyphosis of one or more vertebrae, focal spinal cord narrowing with absent exiting nerve roots, congenital paraplegia and congenital lower limb deformity.<sup>3</sup> Bony defects include dysmorphic, hypoplastic or absent vertebrae, focal spinal canal stenosis and subluxation of the spinal cord.<sup>1</sup> It is often difficult to precisely assess the extent of the anomalous segment because of the pres-



Fig. 1b. Lateral spinal X-ray demonstrates absent T12 and lumbar vertebrae.



Fig. 2b. Axial T2 MRI demonstrates horseshoe kidney (arrowed).

ence of indeterminate, aplastic, hypoplastic or incompletely segmented vertebrae. The condition is segmental with vertebrae above and below the malformation. The spinal cord at the level of the abnormality is typically thinned or indiscernible, with an absence of nerve roots and a bulky, low-lying cord segment present caudal to the focal abnormality in most cases; this is a unique feature of SSD. However, in the presence of a low lesion with lumbosacral spine involvement, the segmental abnormality is too caudal for normal cord to develop below it.<sup>1</sup> In these instances, as demonstrated in our case reports, there is no spinal cord visible distal to the lesion.

## Embryology

Initially, it was believed that the lesion arose during the embryological stage of primary neurulation. Data now suggest that these complex malformations occur sooner, during gastrulation. Gastrulation is the process whereby the bilaminar embryonic disk is converted into a trilaminar disk, which occurs in the third gestational week.<sup>1</sup> There is transient embryonic communication between the amniotic and yolk sacs, disappearing at the beginning of the third week of gestation, just before primary neurulation.<sup>3</sup> Therefore, chordo-mesodermal derangement during gastrulation related to positional programmed cell death of cells with incorrect axial specification can result in SSD.<sup>1</sup> The severity of the disorder correlates with the degree of notochordal cell depletion. From this developmental theory, associated visceral abnormalities can be explained, including neurenteric fistula, as reported by Tortori-Donati *et al.*<sup>1</sup>

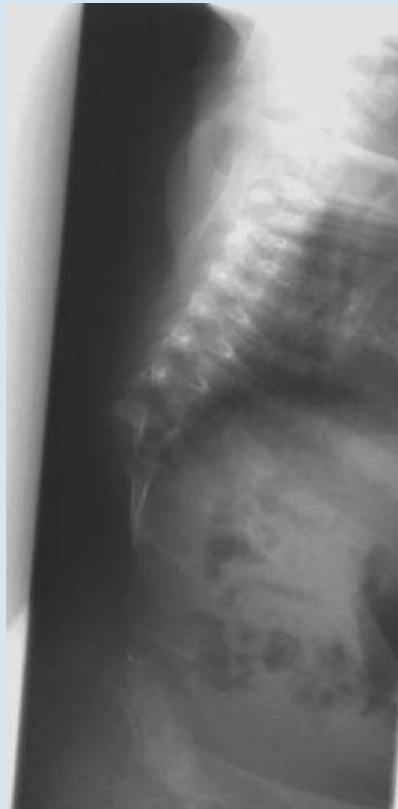
SSD and caudal regression syndrome (CRS) probably represent two faces of a single spectrum of segmental malformations of the spine and spinal cord. They differ from an embryological point of view in the segmental location of the derangement along the longitudinal axis of the embryo. In SSD, the intermediate segment is involved as opposed to the caudal segment in CRS.<sup>1</sup>

## Clinical presentation

Neurological features depend on the severity of the malformation and on the segmental level along the longitudinal embryonic axis, the degree of resulting kyphosis and the presence of associated abnormalities. Bilateral



Fig. 2a. Sagittal T2 MRI demonstrates spinal cord hypoplasia with disconnection of the spinal segments above and below the dysgenetic T12 segment at thoracic level 6, with the bony vertebrae terminating at T10, and complete aplasia of T12. The sacrum and pelvis are normally developed.



*Fig. 3a. Lateral spinal X-ray demonstrating kyphotic deformity at T9 (arrow) where the spinal vertebrae end abruptly.*



*Fig. 3b. AP spinal X-ray demonstrating the agenesis of the lower thoracic, lumbar and sacral spines.*

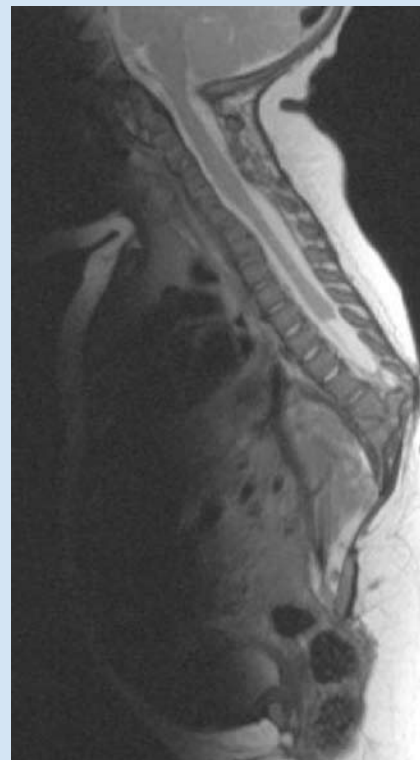
talipes equinovarus deformity of the feet is present in all cases. Patients have hypertrophied, deformed lower limbs. Hip and knee abnormalities have been described. A neurogenic bladder is almost always present and can present as recurrent urinary tract infections complicated by vesico-ureteric reflux and hydronephrosis.<sup>1,4</sup>

### Other associated abnormalities

Closed spinal dysraphisms such as diastomatomyelia, dermal sinus, terminal myelocystocele, lipomas, thickened filum terminale, tethered cord and sacral spina bifida have been described in the literature. Open spinal dysraphisms have not been reported in SSD. Multiple renal abnormalities including crossed ectopia, single kidney and horseshoe kidney are reported, as demonstrated in both our cases. Dextrocardia, situs inversus, tetralogy of Fallot and hypoplasia of the lungs have been described. Bowel abnormalities include imperforate anus, rectovaginal fistulae and malrotation of the bowel.<sup>1,4</sup>

### X-ray features

Moderate to severe kyphosis can be present, with the gibbus apex marking the level of vertebral malformation. Aplastic or hypoplastic vertebrae, hemivertebrae and butterfly vertebrae may be demonstrated at the thoracic, lumbar or sacral regions. There may be marked stenosis or even interruption of the spinal canal at the level of the malformation, and the osseous canal can resemble an hour-glass shape. Associated costal abnormalities, such as bifid, fused, or absent ribs, can also be present.



*Fig. 4a. Sagittal T2 MRI: The thoracic vertebrae and spinal cord terminate at T9. Note the linear bony septum below T9, and the syrinx above.*

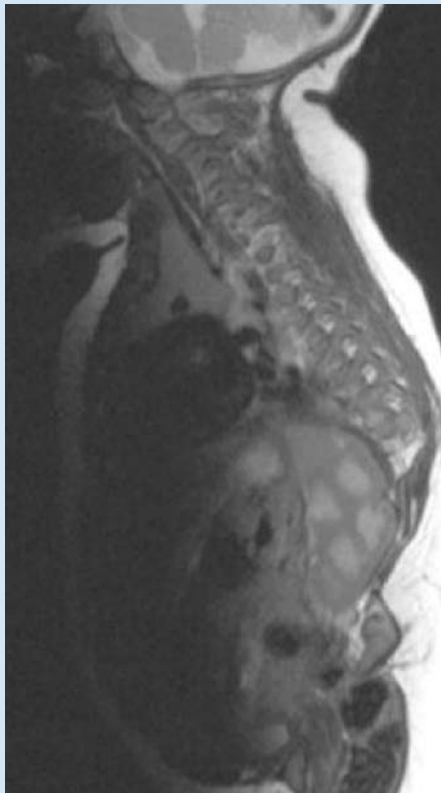


Fig. 4b. Sagittal T2 MRI demonstrating the hydronephrotic kidney.

## Treatment

Even though patients with SSD are not necessarily paraplegic at presentation, they have an increased risk for the development of neurological deficits owing to the associated instability and congenital stenosis of the spine.<sup>1</sup> MRI therefore plays an important role and can influence the surgical strategy.

Hughes *et al.* and Flynn *et al.* (cited in Faciszewski *et al.*)<sup>3</sup> recommend early reconstructive surgery in infancy, so allowing for motor development.<sup>3</sup> Spinal cord decompression and anterior and posterior arthrodesis is pivotal at an early stage to limit the progression of the kyphosis and worsening of neurological impairment.<sup>3,4</sup>

## Conclusion

The SSD malformation is typically segmental with normal vertebrae above or below the anomaly, as if an intermediate segment of the spinal vertebrae and spinal cord were omitted during embryonic development. As there are many associated abnormalities such as neurogenic bladder, renal and other problems, it is important that they are identified early because this aids surgical planning. Imaging therefore plays an important role in the diagnosis, surgical planning and follow-up of such cases.

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