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**Case Report**

# Rare association - type I caudal regression syndrome associated with congenital spinal dermal sinus tract: a multimodality evaluation

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## ABSTRACT

Caudal regression syndrome (CRS) and congenital spinal dermal sinus tract (CSDST) are two complex closed spinal dysraphism without subcutaneous mass. Embryo-pathogenesis behind CRS is deemed to be a fault during gastrulation, however CSDST is a result of defective focal disjunction during primary neurulation. While many associated congenital anomalies have been described with both of these entities, only a few literatures have ever described CRS and CSDST in unison. We present a case with both type 1 CRS with an intramedullary syringohydromyelia and a congenital spinal dermal sinus tract, with their multimodality imaging findings. Prompt identification and accurate diagnosis of CRS is cardinal for initiation of early and appropriate management, before the herald of dreadful complications. CSDST is a remnant tract arising superficially from the skin or subcutaneous plane, extending deep and terminating variably at the spinal cord, meninges or myofascial plane. A newborn's neural system is usually preserved, with usual presentation being with skin stigmata. Nevertheless, this apparently benign sounding entity is associated with various other spinal dysraphisms and predispose the patient to multiple dreadful complications. Thus, a complete evaluation of a dorsal opening is cardinal and a conservative management is discouraged.

**Keywords:** Caudal regression syndrome, Dorsal dermal sinus tract, New associations, Spinal dysraphism

## INTRODUCTION

Spinal dysraphism is a subtype of neural tube defect, that encompass congenital spine and spinal cord developmental disorder occurring due to insult in second to sixth week of intrauterine life, and include incomplete midline closure of nervous, mesenchymal and/or osseous tissue. With an incidence of 1-3 per 1000 live birth, it is second most common birth anomaly. Among the gamut of anomalies under spinal dysraphism, caudal regression syndrome (CRS) and congenital spinal dermal sinus tract (CSDST) are two rare anomalies, classified under complex closed spinal dysraphism without a subcutaneous mass.[1] CRS is an anomaly resulting either from an insult during gastrulation, leading to CRS type 1, or a defective canalization and retrograde differentiation during secondary neurulation, leading to CRS type 2.[2] On the contrary, CSDST arises from a faulty disjunction and focal failure in fusion of surface epithelium during primary neurulation.[3] It is extremely rare to find both of these anomalies occurring in conjunction, and the major reason might be different embryo-pathogenesis of these two entities. In this case report, we are elucidating findings of a patient with CRS type 1 who also had CSDST, among other anomalies.

## CASE REPORT

A seven-year-old male presented to the outpatient department (OPD) with complaints of bladder incontinence and inability to stand. Examination revealed restricted movement of hip, fixed flexion deformity in

bilateral knee and fixed planter flexed bilateral feet. A focal patch of hyperpigmentation was noted in the right para-midline of lower lumbar region; however, no associated subcutaneous mass was noted, as shown in Figure 2e. On X-ray whole spine, there was total sacral and partial lumbar vertebral agenesis with a dysplastic L1 vertebra. Bilateral iliac blades were seen articulating in the midline; however, no articulation was seen with the dysplastic vertebral body, as shown in Figure 1a and b. Considering the complaints, examination findings and X-ray, a diagnosis of caudal regression syndrome was considered. For further evaluation, patient was referred for magnetic resonance imaging (MRI). MRI was acquired with following sequences, T1 and T2 sagittal and axial planes of the whole spine with T2 sagittal brain for screening. MRI revealed an abrupt high blind ending truncated conus medullaris, ending at T8 vertebral level. A thin filum terminale was noted extending from the conus medullaris, coursing caudally and attaching to the anterior dural sac at the level of lower margin of T12 vertebra, as shown in Figure 2a. An intramedullary syringohydromyelia was noted in the lower aspect of the spinal cord, as shown in Figure 2f. A linear T2 hypointense tract was noted arising from the right para-midline subcutaneous plane at the level of T12 vertebral body, coursing caudally to pass through the bifid spinous process of dysplastic L1 vertebral body. A cranial turn was then seen with the tract entering the intradural segment of the spinal canal and seen up-till its attachment to the posterior thecal sac at the level of T12 vertebral body, as shown in Figures 2 b-d.

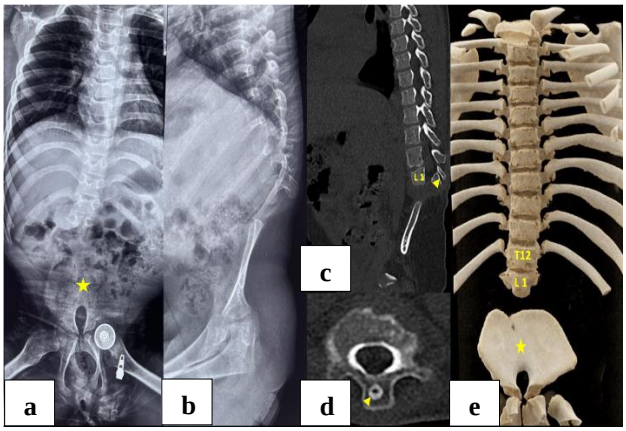


**Figure 1: (a) X-ray AP view whole spine, (b) X-ray lateral view shows total sacral atresia and partial lumber atresia with midline fusion of bilateral iliac blades (star) (shield sign), (c) sagittal, and (d) axial CT images showing dysplastic L1 vertebra with bifid spine (Arrowhead), and (e) virtual reconstructed image of CT scan showing total sacral and L2–L5 vertebrae atresia with midline fusion of bilateral iliac blade (star), with no articulation with the dysplastic L1 vertebra.**

Thus, constituting a "V-shaped" configuration. However, intraspinal segment of the tract could not be resolved by MRI. Thus, confirming a diagnosis of type 1 caudal regression syndrome with congenital spinal dermal sinus tract. A computed tomography (CT) was acquired for assessment of bones, with revealed total agenesis of sacrum, coccyx and lower four lumber vertebra with a dysplastic L1 vertebra. Bilateral iliac blades were seen fused in the midline ("shield sign", however no articulation with the vertebral body was observed, as shown in Figure 1e. L1 vertebra showed bifid spinous process, as shown in Figures 1c and d. There were widely separated pubic symphysis with a distance in-between of 2.3 cm. Our case is classified as type I under Pang's classification and type IV under Ranshaw's classification.

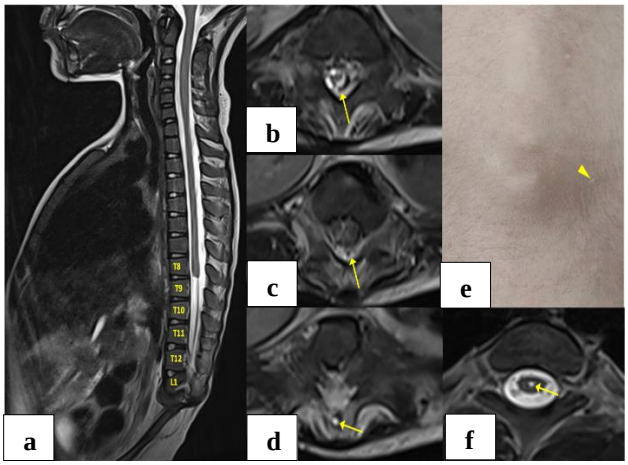


**Figure 2: (a) Sagittal T2 WI MRI of whole spine reveals high blind ending truncated conus medullaris at the level of T8 vertebral body, with a filum (measuring 1.2 mm) extending caudally and attaching to the anterior dural sac at the lower margin of T12 vertebral body; (b-d) axial T2WI MRI images of spine, showing deeper attachment of CSDST to the posterior thecal sac (arrow), its intraspinal course (arrow) and interspinous course (arrow) respectively; (e) gross image of the back of patient showing a skin patch in the right para-midline (arrowhead); and (f) axial T2 WI MRI image showing a central intramedullary syringohydromyelia (arrow).**

## DISCUSSION

CRS, also known as sacral regression syndrome, sacral agenesis or caudal dysplasia sequence, is a complex closed spinal dysraphism characterized by congenital developmental anomalies of caudal part of spine and spinal cord with variable multisystemic involvement, which includes, but is not limited to, nervous, genitourinary, gastrointestinal and skeletal system.[1,2,4] It is associated with other spinal dysraphism, such as myelomeningocele, lipomeningomyelocele, lipoma of spinal cord, low lying cord, cord tethering and diastematomyelia.[2,4] Scarcely in literature, to the best of our knowledge, has it been described with CSDST, which is another rare complex closed spinal dysraphism, consisting of a multilayered epithelial lined tract extending from superficially from

skin or subcutaneous tissue to end deep into the spinal cord, meninges or musculofascial layers.[3] Rarity of this association can be attributed to difference in underlying embryo-pathologies.

Diving in embryology proper, development of spinal cord and its overlying spine is a result of series of interlinked, strictly controlled, orderly occurring intricate processes, which are broadly classified into gastrulation, primary neurulation and secondary neurulation. Type 1 CRS results from a defective gastrulation, because of insult before the fourth week of gestation. They are mostly sporadic, with variable role of genetic and environmental factor as well as embryo vascularization.[2] A correlation between level of spinal cord termination and severity of sacral anomalies suggests a potential association between abnormal cord development with its surrounding tissue.[4] However, CSDST is primarily attributed to defect during primary neurulation. A focal defective fusion of surface epithelium with a complete fusion of underlying neuroepithelium and non-disjunction at this focal point result in CSDST.[3]

The term CRS was first used by Bernard Duhamel in 1964.[2] It is a rare congenital disorder with incidence of approximately 1-3 per 100,000 live births among general population, having equal gender predilection.[5] Various associated risk factors include, uncontrolled maternal gestational diabetes, family or past h/o of CRS and exposure to teratogen in early gestation.[2,5] It has a varied spectrum of presentation, ranging from a low anorectal anomaly (ARM), such as imperforate anus, to sirenomelia (mermaid syndrome). Associated bowel-bladder, renal and musculoskeletal anomalies are usually encountered. Patient usually presents with urinary complaints with or without ARM and walking difficulty.[2,4,5] Many classifications have been proposed over the years, and classification systems by Pang and Ranshaw are widely accepted. Both of these classifications are based on the type of defect and articulation between bones, with few distinctions. Another system of classification divides CRS into type 1 and type 2 based on distinct pattern of spinal cord malformation and associated anomalies. Type 1 CRS, results from a gastrulation defect, having short high riding cord termination and is associated with more anomalies, in contrast to type 2 CRS, which is a secondary neurulation defect, featuring a low-lying cord termination associated with milder anomalies.[4] Prenatal diagnoses by ultrasound and fetal MRI can be made, which aids in the prompt management. Post-nataly, MRI is the mainstay for holistic evaluation of the patient with CRS.[2,4] A multidisciplinary management approach is necessary, with collaboration of radiologist, neurosurgeon, orthopedic surgeon, gastro-surgeon and other specialties, for a prompt and accurate diagnosis, and planning of holistic surgical approach, to provide life-saving treatment in a timely manner and alongside preventing unnecessary surgeries, which in-turn improves patient's quality of life and expectancy.[4,5] While an early response is cardinal for management and good outcome, a diagnostician must rule out CRS mimics, like Currarino sequence; characterized by anorectal atresia or ectopia, coccygeal and partial sacral agenesis, and a pre-sacral mass lesion (anterior meningocele, lipoma or dermoid). Other differentials to be considered are myelomeningocele and cloacal exstrophy.[2,5]

Congenital spinal dermal sinus tract is another, rare closed spinal dysraphism with an incidence of 1 in every 2500 live birth.[6] It is characterized by a squamous epithelium lined tract, arising superficially from the skin or subcutaneous tissue, coursing cranially with a variable deeper extent, ranging from in the spine to myofascial planes.[3,6] Although CSDST can be found anywhere in between occiput to sacrum, it is most commonly reported in lumbar and lumbosacral region.[3,6,7] It has a wide range of presenting features, with most of the new born often having an intact neurological system at birth and presenting with a skin stigmata, which is usually located above intergluteal cleft. These patients can either remain asymptomatic, however most of them develop complaints related to either an associated anomaly or complications.[3,6] Reported associated anomalies include epidermoid, dermoid, tethered cord, split cord malformation and/or syrinx, with only a couple of literature showing association with CRS.[6] Associated complication include acute CNS infection, acute paraplegia, bowel/bladder involvement, sensory deficit and rarely skeletal abnormalities.[6,7] Ackerman and Menezes concluded that delay in diagnosis led to development of neurological complications, which tend to be permanent.[6] Thus, an early diagnosis is key to reducing patient's morbidity. MRI with its superior soft tissue resolution is the imaging modality of choice. One can either find a tubular structure, or occasionally a slender tract with obliterated lumen is observed.[3] However, even an MRI is unable to accurately delineate the intraspinal portion of the tract, and thus operative exploration of all the tract above sacrococcygeal region is advised irrespective of clinical presentation or neuroimaging findings.[6] Owing to the vast associated complications and increased risk of infection, a conservative approach is not taken. While examining a patient with suspected CSDST one should distinguish it from the commoner coccygeal pits/dimple. Coccygeal dimple is a caudally oriented blind ending sinus within intergluteal cleft with no associated cutaneous abnormalities. These tend to be midline, in contrast to CSDST which can be para-midline. Although, coccygeal pit is a benign entity, however few cases with intraspinal extensions have been observed, thus one should investigate a dorsal opening before dismissing it as benign.[6]

## CONCLUSION

Through this manuscript we highlight prime imaging findings in two rare yet important congenital cases of spinal dysraphism, which are seldom reported in unison. Important insight on the embryology, associations, classifications, and management of CRS have been provided, with focus on cardinal role of radiology in early precise diagnosis and classification of disease which facilitates prompt management, resulting in a favourable

prognosis. One must not underscore CSDST, a seemingly benign pathology. It is associated with myriads of other entities. Radiologist hold paramount importance in the diagnosis of CSDST, mapping the tract and in detecting associated complications. Although not the first, we provide excellent multi-modality self-explanatory illustrations of both of these rare entities, aiming to enhance knowledge and reporting in these cases.

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