

# Segmental Spinal Dysgenesis

MOHAMMAD MAMUN-UR-RASHID

## Abstract:

*Report on a rare case of segmental spinal dysgenesis of a 07 months old boy. The boy was born with congenital lower limb deformity and focal kyphosis. Radiological investigations revealed hypoplasia of vertebral bodies and posterior listhesis leading to focal kyphosis. The aim of reporting this case is to highlight the role of radiological investigations in the diagnosis of extent of spinal abnormalities and provide adequate informations for surgical correction.*

**Keywords:** *Segmental spinal dysgenesis, Thoracolumbar kyphosis, Magnetic resonance imaging.*

## Introduction:

Spinal segmental dysgenesis (SSD) is a rare congenital spinal anomaly in which a segment of the spine and spinal cord fails to develop normally with localized agenesis or dysgenesis of the lumbar or thoracolumbar vertebrae with congenital kyphosis. The disease is called segmental because of the presence of normal vertebrae above and below the level of segmentation.<sup>1,2,3,4</sup>

## Case Report:

A 7 months old boy hailing from Rangpur CMH, was referred to radiology department of CMH Dhaka for evaluation of bilateral congenital hydronephrosis, lower limb deformities and focal kyphosis since birth. Detailed history revealed that the boy was delivered by C-Section at 38 weeks of gestation. Prenatal ultrasound revealed bilateral hydronephrosis at 20 weeks and club foot

at 30 weeks. Vertebral anomalies were not document in anomaly scan done at 20 weeks of gestation. At birth he had bilateral clubbed (talipes equinovarus) feet with no history of neurological deficit. His physical examination

demonstrated mild kyphotic deformity (gibbus) at thoracolumbar junction. However, his higher mental functions revealed no retardation. He had a history of recurrent urinary tract infection and persistant electrolyte imbalance. His base line investigations revealed persistent numerous pus cell in urine, urine for C/S shows growth of Klebsiella pneumonia and Pseudomonas species, lymphocytic leukocytosis in CBC. DNA for CMV was detected and IgG for CMV was positive. Echocardiography revealed a moderate ASD. X-Ray dorsolumbar spine demonstrated grade V retrolisthesis. The spine appeared separated with complete offset between upper and lower segments. Abdominal ultrasonography revealed Bilateral hydronephrosis due to PUJ obstruction.

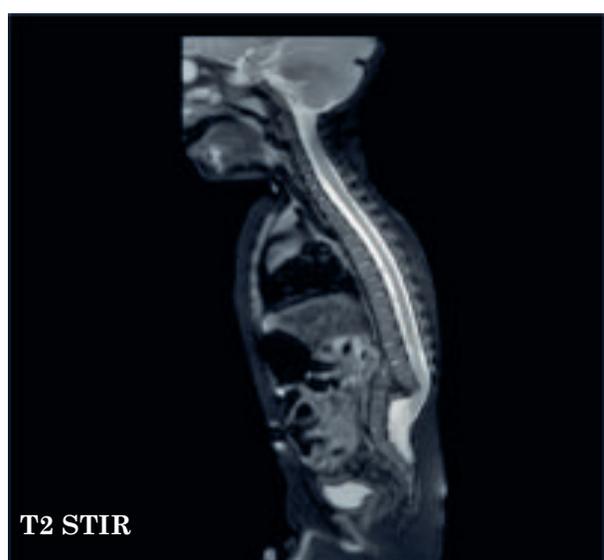
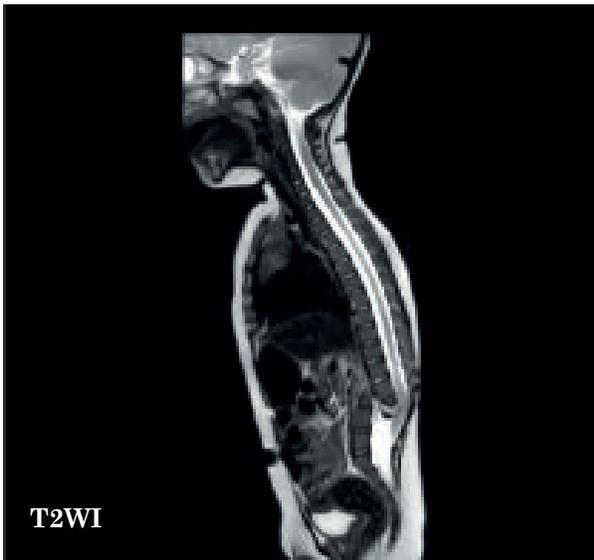
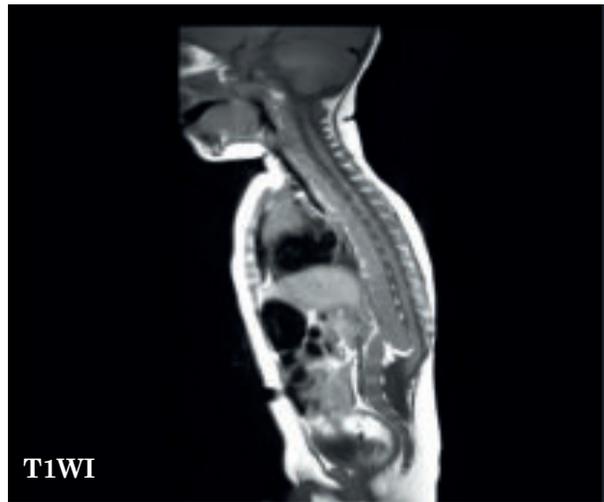
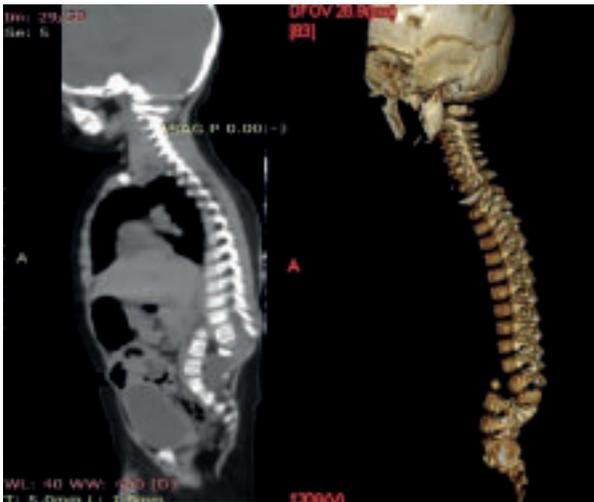
## MRI of dorso-lumbar spine revealed :

- Severe hypoplasia of L2 with absence of its posterior neural element.
- Moderate hypoplasia of L1 and L3 vertebrae with posterior subluxation of L1 vertebra causing subsequent lumbar kyphosis at this level.
- Bulky low lying cord segment is observed just below the level of L3.
- Posteriorly tethered spinal cord adherent to the dura with its distorted morphology.
- The girth of the dorsal portion of spinal cord is mildly narrowed.

**Author of correspondence:** Lt col (Dr) Mohammad Mamun-Ur-Rashid, CI Spl in Radiology & Imaging, CMH Dhaka, Dhaka Cantt Email: drmamun101022@gmail.com

1) Lt col (Dr) Mohammad Mamun-Ur-Rashid, CI Spl in Radiology & Imaging, CMH Dhaka, Dhaka Cantt. Contact Number: 01746652244. Email: drmamun101022@gmail.com

Based upon MRI findings a final diagnosis of segmental spinal dysgenesis was made.



**Discussion:**

Segmental spinal dysgenesis is a malformation that is probably the result of embryonic segmental malformation or focal injury to the developing spine in utero. In utero detection is possible, however affected children are typically identified at the time of birth by a kyphotic deformity (often sharply angled), anomalies in the lower limb (usually equinovarus deformity of the feet and flexion contractures of the hips and knee), hyperreflexia of the lower extremities and bladder dysfunction. Skin overlying the deformity may be bluish.

Imaging reveals a marked focal hypoplasia of the vertebral column, thecal sac and spinal cord, typically at the thoraco lumbar junction. Plain films show a kyphotic deformity with hypoplasia or absence of one or more vertebral bodies. Myelography shows smooth tapering of the thecal sac and spinal cord with a complete or near complete block of contrast at the most severely affected levels. Caudal to the narrowing a round or oval intradural mass represents the lower segment of the spinal cord. CT myelography shows marked narrowing of the bony narrowing of the spinal canal with small remnant of subdural space running through the extra spinal soft tissues. MR may shows variable degree of tapering of the spinal cord to a point of marked narrowing or complete focal absence of the cord. However in most cases, the termination of the upper segment of the spinal cord is abrupt and in some cases it

appears blunted. Both these appearances are similar to the appearance of the spinal cord in patients with caudal regression / lumbosacral hypogenesis. Below the segmental agenesis, the bony spinal canal, thecal sac and spinal cord resume a normal appearance. Evaluation of entire spinal column is essential as affected patients may have associated lipoma, dermal sinuses or hydromyelia.<sup>5</sup>

**References:**

1. Mahomed N & Naidoo J. Spinal Segmental Dysgenesis. *S Afr J Radiol*. 2009;13(2):29. doi:10.4102/sajr.v13i2.542
2. Naik S, Bhoi S, Panigrahi K, Deep N. Segmental Spinal Dysgenesis: A Rare Congenital Spinal Malformation. *Indian J Radiol Imaging*. 2019;29(4):480-1. doi:10.4103/ijri.IJRI\_195\_19 - Pubmed
3. Knafelz E, Lall N, Love M, Bui C, Steven A. Segmental Spinal Dysgenesis. *Ochsner J*. 2020;20(3):244-7. doi:10.31486/toj.19.0094 - Pubmed
4. Paolo Tortori-Donati, Maria Paola Fondelli, Andrea Rossi, Charles A. Raybaud, Armando Cama, Valeria Capra. Segmental Spinal Dysgenesis: Neuroradiologic Findings with Clinical and Embryologic Correlation. *AJNR Am J Neuroradiol*. 1999;20(3):445-56. - Pubmed
5. A. James Barkovich, Charles raybaud, *Pediatric neuroimaging*, fifth edition.