ScientificScholar®
Knowledge is power
Publisher of Scientific Journals

www.surgicalneurologyint.com

Surgical Neurology International

Editor-in-Chief: Nancy E. Epstein, MD, Clinical Professor of Neurological Surgery, School of Medicine, State U. of NY at Stony Brook.

SNI: Pediatric Neurosurgery

Editor
Frank Van Calenbergh, MD
University Hospitals; Leuven, Belgium

SNI
Open Access

*Case Report*

# Severe type of segmental spinal dysgenesis with complete disconnection of the spinal cord and vertebra associated with open neural tube defect

Ai Kurogi[1], Nobuya Murakami[1], Takafumi Shimogawa[2], Nobutaka Mukae[2], Satoshi O. Suzuki[3], Toru Yamaguchi[4], Koji Yoshimoto[2], Takato Morioka[5]

[1]Department of Neurosurgery, Fukuoka Children's Hospital, [2]Department of Neurosurgery, Kyushu University, [3]Department of Psychiatry, Shourai Hospital, [4]Department of Orthopedic Surgery, Fukuoka Children's Hospital, [5]Department of Neurosurgery, Harasanshin Hospital, Fukuoka, Japan.

E-mail: *Ai Kurogi - aironron8282@gmail.com; Nobuya Murakami - murakami.n@fcho.jp; Takafumi Shimogawa - shimogawa28@gmail.com; Nobutaka Mukae - mukaen0203@gmail.com; Satoshi O. Suzuki - sosuzuki@shouraikai.jp; Toru Yamaguchi - yamaguchi.to@fcho.jp; Koji Yoshimoto - yoshimoto.koji.315@m.kyushu-u.ac.jp; Takato Morioka - takato@ns.med.kyushu-u.ac.jp

***Corresponding author:**
Ai Kurogi,
Department of Neurosurgery,
Fukuoka Children's Hospital,
Fukuoka, Japan.

aironron8282@gmail.com

Received : 14 February 2023
Accepted : 05 April 2023
Published : 28 April 2023

**DOI**
10.25259/SNI_156_2023

**Quick Response Code:**

## ABSTRACT

**Background:** Severe type of segmental spinal dysgenesis (SSD) is a rare and complex anomaly in which the spinal cord completely disconnects at the portion of the spinal dysgenesis. Although closed spinal dysraphisms have been associated with SSD, to the best of our knowledge, the association between open neural tube defect (ONTD) and SSD is significantly rare, with only one case being reported to date.

**Case Description:** We report a case of an infant with severe SSD and a disconnected spinal cord and spinal column at the thoracolumbar junction associated with myelomeningocele (MMC) in the lumbosacral region. The patient presented severe neurological deficits in the legs and impaired bowel function. The spinal column of L1–L3 was absent. The lower spinal segment consisted of neural placode at the L5–S1 level and no connecting structure between the upper and lower spinal cords. A repair surgery for MMC, including cord untethering and dura plasty, was performed. Histopathological findings revealed a neural placode consisting of a neuroglial tissue and leptomeninges.

**Conclusion:** The management of severe SSD during the perinatal period is more challenging when it is associated with ONTD. We report detailed neuroradiological, intraoperative, and histological findings of such a case and discuss the embryopathogenesis of the associated ONTD and the treatment strategies.

**Keywords:** Gastrulation, Junctional neural tube defect, Neurulation failure, Open neural tube defect, Segmental spinal dysgenesis

## INTRODUCTION

Segmental spinal dysgenesis (SSD) is a rare congenital abnormality in which a segment of the spine and spinal cord fails to develop appropriately, and in most severe cases, the spinal cord completely disconnects at the portion of the spinal dysgenesis, presenting with severe neurological deficits.[3,15,21] SSD has been hypothesized to be caused by notochord malformation disorders during gastrulation, which inhibits the subsequent formation of both the spinal vertebrae and neural tube, resulting in the segmental defect of the spinal column and spinal

cord.[3,22] Although closed spinal dysraphisms such as tight filum terminale, filar lipoma, and diastematomyelia have been associated with SSD,[3,22] to the best of our knowledge, the association between SSD and open neural tube defect (ONTD) has been described in only one case of SSD.[15] In that case, magnetic resonance imaging (MRI) confirmed SSD associated with myelomeningocele (MMC) and intracranial Chiari II features; the authors did not consider the reason for the coexistence SSD and MMC.[15]

Herein, we present detailed neuroradiological, intraoperative, and histological findings of the case of an infant with severe SSD and a disconnected spinal cord and spinal column at the thoracolumbar junction associated with MMC in the lumbosacral region.

## CASE REPORT

A 34-year-old primigravida woman was referred to our hospital as no movements were detected in the fetus's legs on transabdominal ultrasound examination at 26 + 5 weeks of gestation. Prenatal MRI performed at 27 + 4 weeks of gestation failed to reveal apparent abnormalities in the lumbosacral regions, hydrocephalus, or a Chiari malformation.

The mother vaginally delivered a girl weighing 2,471 g and with an Apgar score of 8 at 37 + 4 weeks of gestation. A skin defect (15 × 25 mm) associated with a dysplastic neural placode exposed to the surface was detected in the infant at the lumbosacral region without cerebrospinal fluid (CSF) leakage [Figure 1a-1]. The infant had spastic paraplegia and severe flexor contractures in the cross-legged position [Figure 1a-2], and the infant's anus was patulous with fecal incontinence. The infant had urinated frequently, indicating urinary dysfunction.

Spinal three-dimensional (3D) computed tomography revealed that the spinal column at L1–L3, including both vertebrae and posterior elements, was absent and that the lumbar spine below L4 and bifid sacrum deviated anteriorly [Figures 1b-1 and b-2]. The spine showed no other deformities. Postnatal MRI examinations, including 3D-heavily T2-weighted images and 3D-T1-weighted images,[11,12] showed that the spinal cord was divided into two segments above and below the spinal agenesis [Figures 1c and d, Figure 2a]. The upper segment seemed normal [Figure 2b-1], but the spinal cord abruptly ended with a "cigar-shaped" appearance at the lower thoracic and caudal regions [Figures 1c and d, Figures 2b-2 and b-3]. The lower segment originated from around the L4 level, of which caudal end was a neural placode without cystic component at the S1 level [Figure 2b-4]. These features were consistent with MMC. Neither band-like structures nor delineation of the intensity of CSF was detected between the upper and lower spinal cords [Figures 2b-2 and b-3]. The brain MRI did not provide evidence of hydrocephalus or a Chiari malformation Type II.

On the day after birth, the infant underwent repair surgery for MMC. The open neural placode was circumferentially resected from the surrounding skin, and proximal spinal cord and nerve roots continuing with the ventral side of the neural placode were observed in the open dural sac filled with CSF [Figure 3a]. No pulsation of the CSF was observed and the Valsalva maneuver did not increase the CSF volume. The proximal depth of the spinal canal could not be clearly visualized. Direct stimulation of the placode, proximal spinal cord, and nerve roots elicited no evoked compound muscle action potentials on intraoperative neurophysiological monitoring of the legs and anus (maximum current, 30 mA). We resected the surrounding margin of the neural placode and approximated with pia-arachnoid sutures, followed by multiple-layer closure, including the dura mater and muscle layer.[18] Postoperatively, the patient's neurological function did not change from the preoperative level. At 3 months, she presented with mild hydronephrosis detected on ultrasonography.

The histopathological examination of the resected tissue from the margin of the neural placode showed a glial fibrillary acidic protein-immunopositive neuroglial tissue adjoining a fibrotic leptomeningeal tissue [Figures 3b and c]. The histopathological findings confirmed the diagnosis of MMC.

## DISCUSSION

The embryopathogenesis of SSD has been hypothesized to be the positioning error of the chordomesoderm cells, which is the mesoderm that gives rise to the notochord. The error causes apoptosis of the cells in the malpositioned segment (positional apoptosis) during gastrulation.[21,22] As the chordomesoderm cells act as an inducer of neural plate and somites,[6] the error leads to a dysgenesis of spinal cord and spinal vertebral bodies at the pathological segment in severe cases of SSD.[3,15,21] The notochordal process is thought to elongate caudally of the malpositioned segment by adding the chordomesoderm cells at its caudal end;[17] in other words, the primary neural tube formation is allowed to continue caudally and develop to the lower spinal segment in SSD cases.[22] However, the lower spinal segment is low lying and thickened in most cases of SSD, of which in embryogenesis, it has been postulated that the increased number of chordomesoderm cells, which may have been prevented from moving and dammed at the malformed segment and evaded positional apoptosis, forms the thicker spinal cord.[3,21,22]

Although rare coexistence of ONTD in SSD cases has been explained by the findings of studies on avian embryos demonstrating that removal of the notochordal segment does not affect the formation of dermatome and

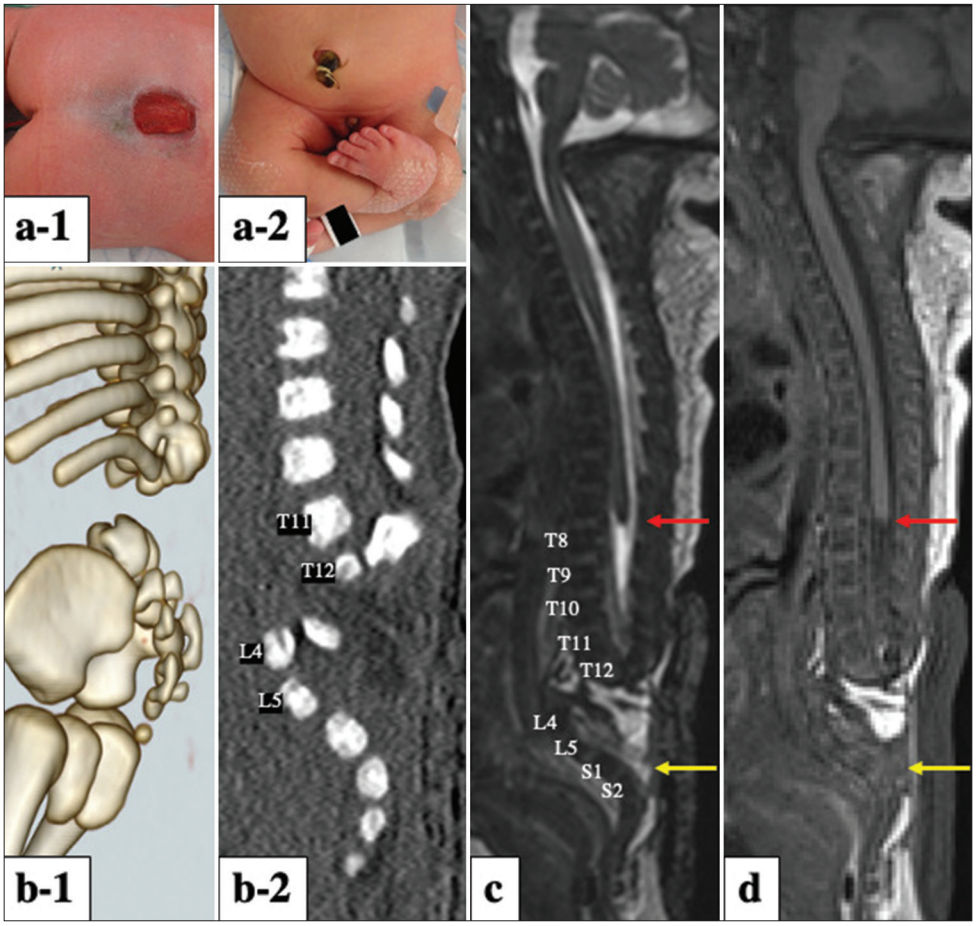


**Figure 1:** Photographs at birth showing (a-1) a neural placode at the lumbosacral region with an open skin defect (15 × 25 mm) exposed to the surface and (a-2) the patient's lower limbs in the cross-legged position with severe flexor contractures. (b-1) Three-dimensional and (b-2) sagittal spinal computed tomography images showing a complete absence of the L1–L3 lumbar vertebrae and dislocation of the lower segment, which deviates anteriorly at the level of the absence. (c) Sagittal three-dimensional-heavily T2-weighted and (d) sagittal T1-weighted spoiled gradient-recalled echo images also demonstrated two completely separated spinal cord segments. The upper segment ends at T8, with a "cigar-shaped" appearance instead of the usual tapered conus (red arrow). The lower segment is located from L4 to S1, displaying an exposed placode (yellow arrow) lying flush with the skin surface with an underlying subarachnoid space, consistent with myelomeningocele.

myotome (future skin and muscle, respectively) but causes defects of sclerotome,[13,22] the exact mechanism remains uncertain. The present case had MMC, which was located caudal to the segmental dysgenesis. Considering that the embryopathogenesis of MMC is the primary neurulation failure,[16,18] if the abovementioned chordomesoderm cells cannot properly act as a neural inducer at the lower segment, the primary neural tube may not have closed properly, resulting in an ONTD in the present case.

The morphological feature of SSD in the aspect of the disconnected upper and lower spinal cords has been advocated to be similar to that of a new disease entity, called a junctional neural tube defect (JNTD).[24] JNTD appears during the short period between the end of primary neurulation and the beginning of secondary neurulation with morphological feature of a nonfunctioning band-like structure connecting the upper and lower cords.[1,4,7,8,10,20,23,24] Although the vertebral body is relatively spared, the posterior elements of the spine are mostly involved, such as hemivertebrae or a vertebral arch defect.[14] The present case was not likely to be JNTD based on the following morphological findings: (1) the absence of a nonfunctional band connecting the upper and lower cords, which has been demonstrated in all previously reported cases of JNTD, and (2) a complete absence of the vertebral body and the posterior element of L1–L3, which may be due to the error that occurred around the same time as neural tube. Voiding of frequent urine in the present

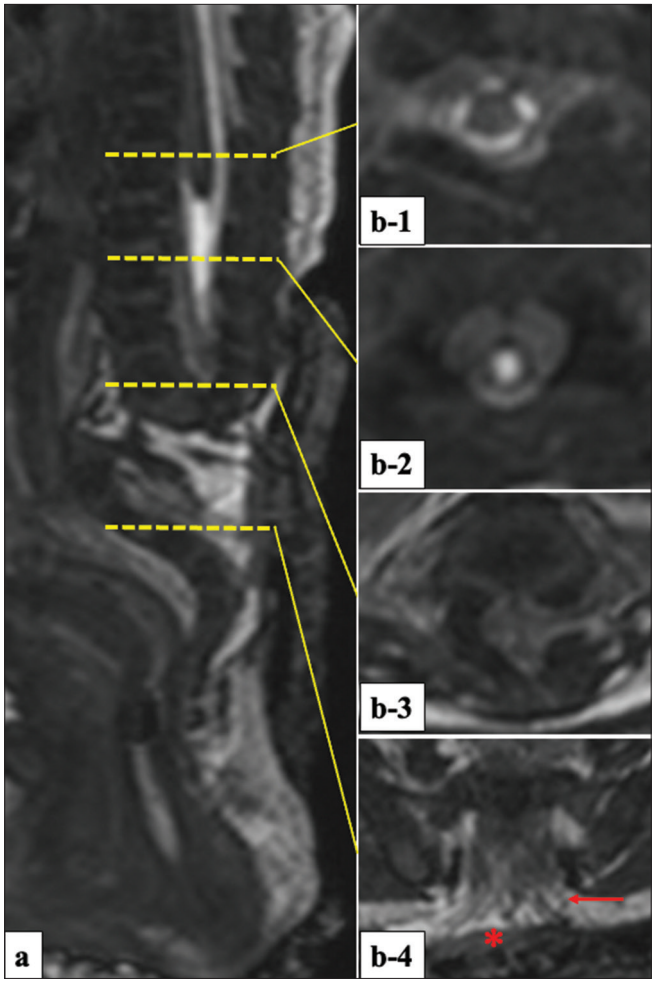


**Figure 2:** A sagittal view (a) and serial axial views (b-1-4) of 3D-hT2WI delineating detailed structures. (b-1) Normal appearance of upper spinal cord at T7 level. (b-2) Absence of band-like structure caudally to the upper spinal cord. (b-3) No signal intensity of CSF at the portion between the upper and lower dural sac. (b-4) Bundle of nerve roots (red arrow) in the subarachnoid space and the neural placode (red asterisk) attached to the skin surface. 3D-hT2WI: Three-dimensional heavily T2-weighted imaging, CSF: Cerebrospinal fluid.

case is assumed to be dependent on a primitive segmental spinal micturition reflexes in the sacral spinal cord, with which humans are naturally born. In normal individuals, the supraspinal pathways emerge during postnatal development, and the segmental reflex diminishes and remains suppressed. However, in the present case, achieving the developed form of voiding was difficult because of the disconnection of the spinal cord, such as in patients with chronic spinal cord injuries.[5]

Considering that most cases of SSD have severe bladder dysfunction as they age,[3,22] the patient's dysuria may worsen in the near future, and a close follow-up with periodical urological examinations is mandatory.

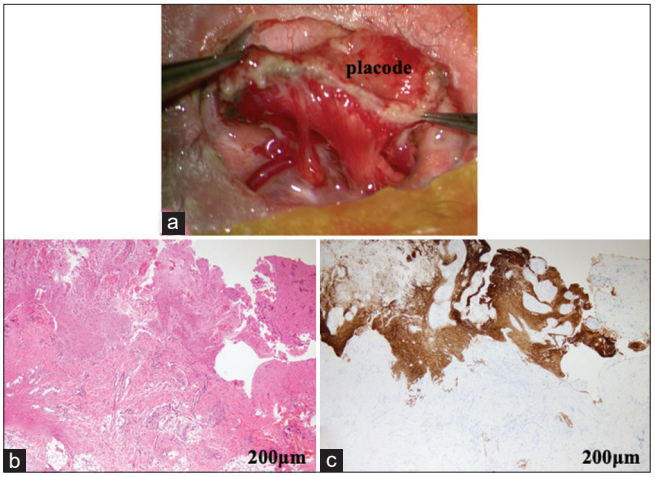


**Figure 3:** (a) An intraoperative picture of the neural placode resected from the attached skin, demonstrating proximal spinal cord and nerve roots arising from the dorsal aspect of the placode. Histopathological findings of the resected margin of the neural placode stained with hematoxylin and eosin (b) and immunostained for glial fibrillary acidic proteins (GFAP, c). The specimen comprises a GFAP-immunopositive neuroglial tissue including mature neurons and an adjoining fibrotic leptomeningeal tissue.

In the present case, we performed dura plasty for the MMC immediately after birth to prevent infection. Regarding cord tethering, severe cases of SSD were unlikely to benefit from untethering surgery because the neurological disturbances appeared to be related to congenital hypoplasia of the nerve roots and spinal cord, and none of the studies have reported progressive neurological deficits during the follow-up periods.[2,9,19]

Patients with SSD require orthopedic treatment to establish and maintain spinal stability and to slow the progressive worsening of neurological functioning due to kyphosis.[2,9,22] Our approach, in this case, includes rehabilitation, in the first few years, to increase the patient's range of motion of her lower extremities and enable diaper and clothing changes. Subsequently, we plan to perform orthopedic surgery, although multiple surgical interventions may be required, considering the fragile bone formation and inadequate fixation.[2,9,19]

## CONCLUSION

Severe SSD has a complex and serious abnormality and earlier neurosurgical intervention is required during the perinatal period in cases associated with ONTD. In the future, studies including a large number of patients are warranted for further understanding of this congenital disorder.

### Acknowledgment

We would like to thank Drs. Tadamune Kinjyo and Naoyuki Nakanami, Departments of Neonatology and Obstetrics, respectively, for supporting our study.

### Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

### Financial support and sponsorship

Research Foundation of Fukuoka Children's Hospital.

### Conflicts of interest

There are no conflicts of interest.

## REFERENCES

1. Ali M, McNeely PD. Junctional neural tube defect: A supporting case report. Childs Nerv Syst 2018;34:1447-8.
2. Bristol RE, Theodore N, Rekate HL. Segmental spinal dysgenesis: Report of four cases and proposed management strategy. Childs Nerv Syst 2007;23:359-64.
3. Chellathurai A, Ayyamperumal B, Thirumaran R, Kathirvelu G, Muthaiyan P, Kannappan S. Segmental spinal dysgenesis-"redefined." Asian Spine J 2019;13:189-97.
4. Dady A, Havis E, Escriou V, Catala M, Duband JL. Junctional neurulation: A unique developmental program shaping a discrete region of the spinal cord highly susceptible to neural tube defects. J Neurosci 2014;34:13208-21.
5. De Groat WC, Griffiths D, Yoshimura N. Neural control of the lower urinary tract. Compr Physiol 2015;5:327-96.
6. Dias MS, Walker ML. The embryogenesis of complex dysraphic malformations: A disorder of gastrulation? Pediatr Neurosurg 1992;18:229-53.
7. Eibach S, Moes G, Hou YJ, Zovickian J, Pang D. Unjoined primary and secondary neural tubes: Junctional neural tube defect, a new form of spinal dysraphism caused by disturbance of junctional neurulation. Childs Nerv Syst 2017;33:1633-47.
8. Eibach S, Pang D. Junctional neural tube defect. J Korean Neurosurg Soc 2020;63:327-37.
9. Faciszewski T, Winter RB, Lonstein JE, Sane S, Erickson D. Segmental spinal dysgenesis: A disorder different from spinal agenesis. J Bone Joint Surg 1995;77:530-7.
10. Florea SM, Faure A, Brunel H, Girard N, Scavarda D. A case of junctional neural tube defect associated with a lipoma of the filum terminale: A new subtype of junctional neural tube defect? J Neurosurg Pediatr 2018;21:601-5.
11. Hashiguchi K, Morioka T, Murakami N, Yamashita K, Hiwatashi A, Ochiai M, *et al*. Clinical significance of prenatal and postnatal heavily T2-weighted magnetic resonance images in patients with myelomeningocele. Pediatr Neurosurg 2015;50:310-20.
12. Hashiguchi K, Morioka T, Yoshida F, Miyagi Y, Mihara F, Yoshiura T, *et al*. Feasibility and limitation of constructive interference in steady-state (CISS) MR imaging in neonates with lumbosacral myeloschisis. Neuroradiology 2007;49:579-85.
13. Hirano S, Hirako R, Kajita N, Norita M. Morphological analysis of the role of the neural tube and notochord in the development of somites. Anat Embryol (Berl) 1995;192:445-57.
14. Kaplan KM, Spivak JM, Bendo JA. Embryology of the spine and associated congenital abnormalities. Spine J 2005;5:564-76.
15. Kashgari AA. Segmental spinal dysgenesis with open spinal dysraphism and Chiari II features, case report. Radiol Case Rep 2020;15:1965-7.
16. Marín-Padilla M. Cephalic axial skeletal-neural dysraphic disorders: Embryology and pathology. Can J Neurol Sci 1991;18:153-69.
17. Moore K, Persaud TV, Torchia M. The Developing Human: Clinically Oriented Embryology. 11th ed. Philadelphia, PA: Saunders; 2018.
18. Morioka T, Hashiguchi K, Mukae N, Sayama T, Sasaki T. Neurosurgical management of patients with lumbosacral myeloschisis. Neurol Med Chir 2010;50:870-6.
19. Pavlova OM, Ryabykh SO, Kozyrev DA, Gubin AV. Surgical treatment of thoracolumbar segmental spinal dysgenesis: Optimal type of fusion. World Neurosurg 2017;106:551-6.
20. Schmidt C, Voin V, Iwanaga J, Alonso F, Oskouian RJ, Topale N, *et al*. Junctional neural tube defect in a newborn: Report of a fourth case. Childs Nerv Syst 2017;33:873-5.
21. Scott RM, Wolpert SM, Bartoshesky LE, Zimbler S, Karlin L. Segmental spinal dysgenesis. Neurosurgery 1988;22:739-44.
22. Tortori-Donati P, Fondelli MP, Rossi A, Raybaud CA, Cama A, Capra V. Segmental spinal dysgenesis: Neuroradiologic findings with clinical and embryologic correlation. Am J Neuroradiol 1999;20:445-56.
23. Tortori-Donati P, Rossi A, Cama A. Spinal dysraphism: A review of neuroradiological features with embryological correlations and proposal for a new classification. Neuroradiology 2000;42:471-91.
24. Wang KC, Lee JS, Kim K, Im YJ, Park K, Kim KH, *et al*. Do junctional neural tube defect and segmental spinal dysgenesis have the same pathoembryological background? Childs Nerv Syst 2020;36:241-50.

**How to cite this article:** Kurogi A, Murakami N, Shimogawa T, Mukae N, Suzuki SO, Yamaguchi T, *et al*. Severe type of segmental spinal dysgenesis with complete disconnection of the spinal cord and vertebra associated with open neural tube defect. Surg Neurol Int 2023;14:149.