Letter to the Editor

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Junctional Neural Tube Defect : Two Case Report

Bin Yuan,[1] Shungen Huang,[2] Xiangming Yan,[3] Hangzhou Wang[1]

Departments of Pediatric Neurosurgery,[1] Children's Hospital of Soochow University, Suzhou, China

Departments of Pediatric General Surgery,[2] Children's Hospital of Soochow University, Suzhou, China

Departments of Pediatric Urology,[3] Children's Hospital of Soochow University, Suzhou, China

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Address for correspondence : **Hangzhou Wang**

Department of Neurosurgery, Children's Hospital of Soochow University, 92 Zhongnan Street, Suzhou 215006, China

Tel : +86-, Fax : +86-, E-mail : 18896544006@163.com, ORCID : https://orcid.org/

To the Editor,

In 2022-2023, we encountered two clinical cases, and after reviewing the literature, we found that the disease belongs to the new spinal dysraphic malformation: junctional neural tube defect(JNTD)[3]. In the previous literature,we found only six reported cases. Here we present the data of two patients in our center in hope to provoke further research to obtain deeper insights into JNTD.

Case 1

The first case concerned a two-year-old boy who had had a recurrent infection in the sacrococcygeal fossa for two years. The patient also exhibited several malformations. Diagnosed at birth with unilateral equinovarus, he underwent surgery for the defect, and was immobilized in plaster splints. Moreover, he had unilateral hydronephrosis, for which he underwent pyeloureteroplasty at the age of eight months. Examination of his lower extremities showed the deep tendon reflexes(Patellar and Achilles) were normal. Motor testing showed unilateral of the extensor digitorum longus, extensor hallucis longus, and gastrognemius muscles were weakness. The tone of anus and the response of bulbocavernous reflex were normal. But this patient did not have an EMG.

We began by administering anti-infective treatment and performed an MRI scan which revealed a blunt spinal cord at level T12 and an unidentifiable structure resembling a cone at level S1/2 connected to the upper spinal cord by a fibrous band(Fig. 1) and the urodynamic study revealed detrusor hyperreflexia，small capacity and heavily trabeculated bladder. Given

the recurrent sacrococcygeal infection and the abnormal MRI finding, we recommended surgical treatment and the pre-operative diagnosis was sinuses with infection. The goal of surgery is to remove the sinuses that are the source of the infection, and the abnormal tissues in the spinal canal were also investigated. Intraoperatively, we found a sinus tract extending from the subcutis to the dura. Another finding was the unidentifiable structure at S1/2 that clearly appeared on MRI, and it was determined to be a conus and fibrosis of filum terminale that was cut intraoperatively. Unfortunately, the small surgical incision which exposed the upper spinal cord opposite L4 down to the caudal end of the lower spinal cord at S2 made it impossible for us to monitor a large number of nerves. And the histopathological of the sinus tract revealed inflammatory sinuses lined with stratified squamous epithelia. One year after surgery, the boy was declared cured of the recurrent infections; however, the urinary, bowel, and lower limb-related problems persisted, and MRI also showed no change in the position of the conus(Fig. 2).

Case 2

The second case we reported was a five-year-old boy with lower limb problems(clubfoot). When the boy was aged three months, his left foot was operated on for the congenital defect and was held in a cast for a long time. Unfortunately, the clubfoot did not improve until he was one year old. Consequently, his neurosurgeon considered performing an MRI scan. The MRI scans showed the lipomatous filum terminale, which his doctor decided to surgically remove.

The outpatient physical examination showed atrophy of the left lower extremity and a

clubfoot, but both feet were with no plantarflexion or dorsiflexion bilaterally. The patellar reflex was normal and the Achilles reflex was weakness. He also did not have an EMG. There were no sacrococcygeal pits or cutaneous stigmata implying occult spinal dysraphism. Anal and bulbocavernosus reflexes were normal. Urodynamic study and urine ultrasound showed no signs of uropathy. The patient requested a repeat MRI and a treatment recommendation. Interestingly, MRI showed a discontinuous lumbar spinal cord(Fig. 3). Further, the three segments of this “upper” part of the spinal cord were found to comprise an enlarged entrance canal. The conus medullaris was visualized in the “lower” part of the spinal cord at the L4 vertebral column. As in case 1, the upper spinal cord and the lower spinal cord were connected by a fibrous cord. We recommended follow-up treatment and rehabilitation of the lower limbs. Regrettably, the loss of data made it impossible for us to obtain a preoperative MRI scan.

In 2014, Dady et al. [2] first introduced the concept of junctional neurulation as a process occurring between the end of primary neurulation and the beginning of secondary neurulation. The authors showed that this process is distinct from primary neurulation and secondary neurulation by describing its underlying mechanisms.

Junctional neural tube defect (JNTD) was first described in 2016 by Eibach et al. [3] using three patients who had two separate spinal cord segments that appeared to be functionally unconnected and physically separated, yet, connected by a bright band on MRI. In the years that followed, Schmidt et al. [4], Florea et al. [5], and Ali et al. [6] each reported one such case. Eibach and Pang [7] classified JNTD as a rare and relatively new dysraphic malformation of the spine affecting embryologic processes. The authors provided a detailed

description of the clinical manifestation, imaging features, and electrophysiologic characteristics, hypothesizing the embryogenetic mechanism of JNTD. Currently, there is no consensus on the treatment of JNTD, and urinary system control and lower limb rehabilitation are particularly important in this respect.

Despite their shared similarities, JNTD and segmental spinal dysgenesis(SSD) are fundamentally different. SSD can affect any spinal segment, whereas JNTD only affects the spinal cord segment between the primary and secondary neural tubes[7]. Another difference is the fact that in SSD there is a spinal deformity compressing the corresponding spinal cord, which is not the case in JNTD [7].

The two cases we presented are consistent with JNTD in terms of both clinical presentation and magnetic resonance imaging. Hence, we believe that our patients should be included in this category.

AUTHORS' DECLARATION

Conflicts of interest

No potential conflict of interest relevant to this article was reported.

Informed consent

This type of study does not require informed consent.

Data sharing

None

Preprint

None

ORCID

Bin Yuan https://orcid.org/0009-0002-9894-663X

Shungen Huang https://orcid.org/

Xiangming Yan https://orcid.org/

Hangzhou Wang https://orcid.org/

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Figure legend

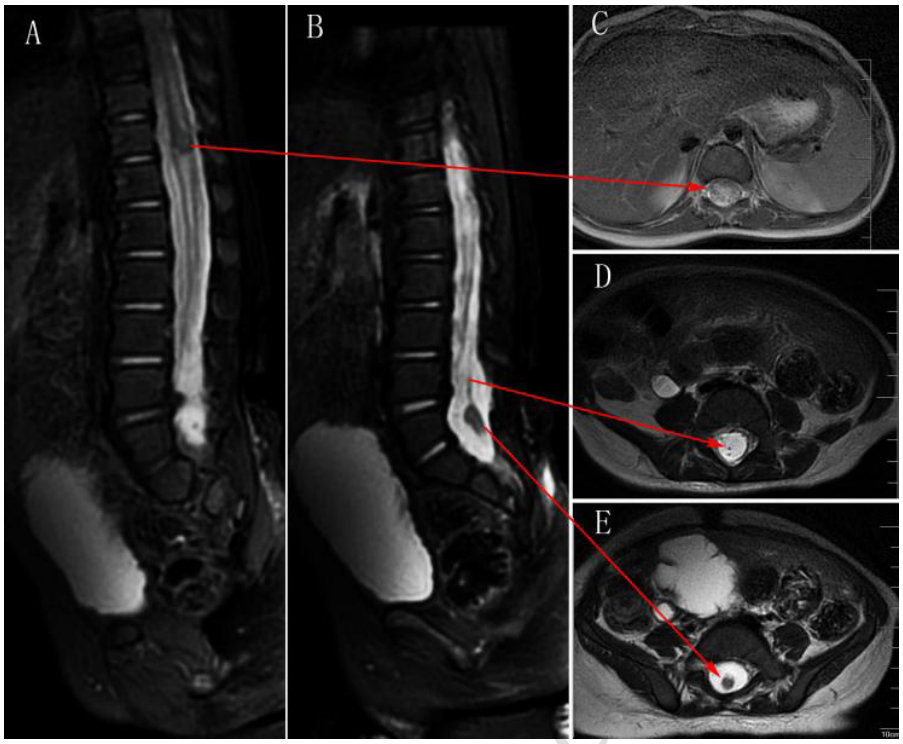

Fig. 1. Preoperative T2-weighted MRI of patient 1. A : Sagittal view shows a blunt-end upper spinal cord at T_{12}/L_1. B : Sagittal view shows the lower spinal cord liking a conus at $S_{1/2}$. C : Axial view at T_{12} shows the upper spinal cord. D : Axial view at L_4 shows the connecting fibrous band between the upper spinal cord and the lower spinal cord. E : Axial view shows the lower spinal cord.

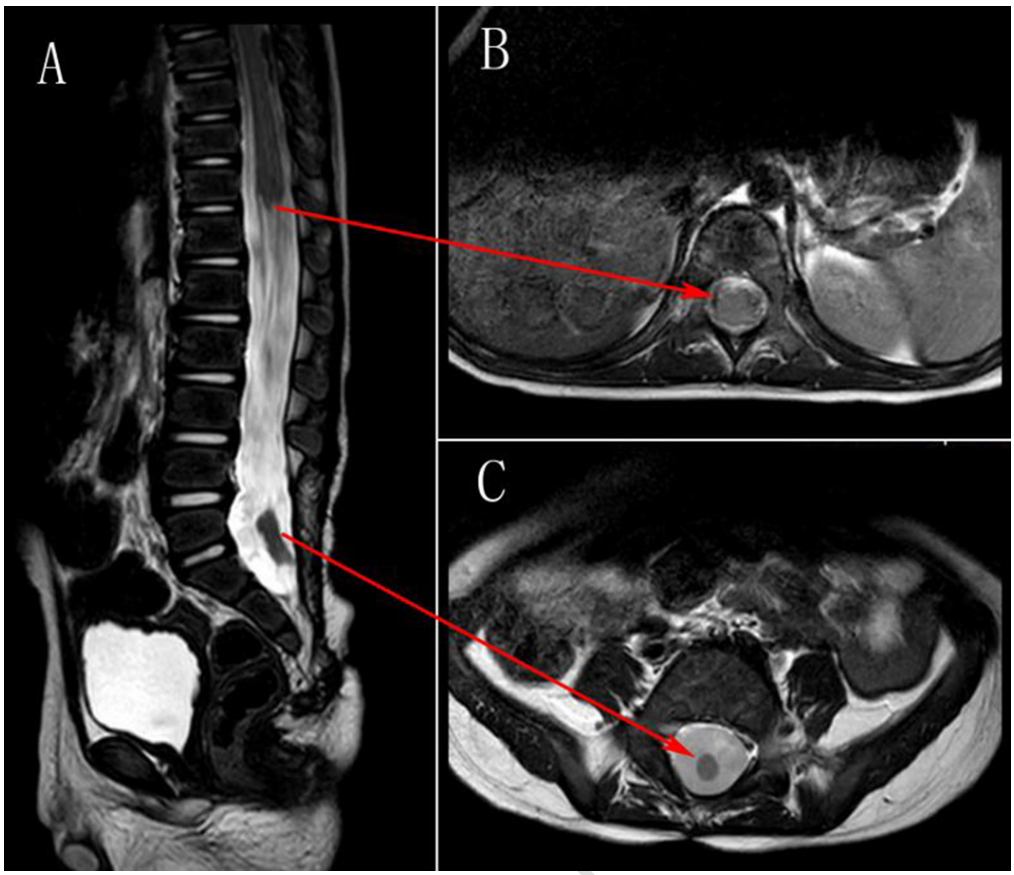

Fig. 2. T2-weighted MRI of patient 1 one year after surgery. A : Sagittal view shows the upper spinal cord at T_{12}/L_1 and the lower spinal cord at $S_{1/2}$.

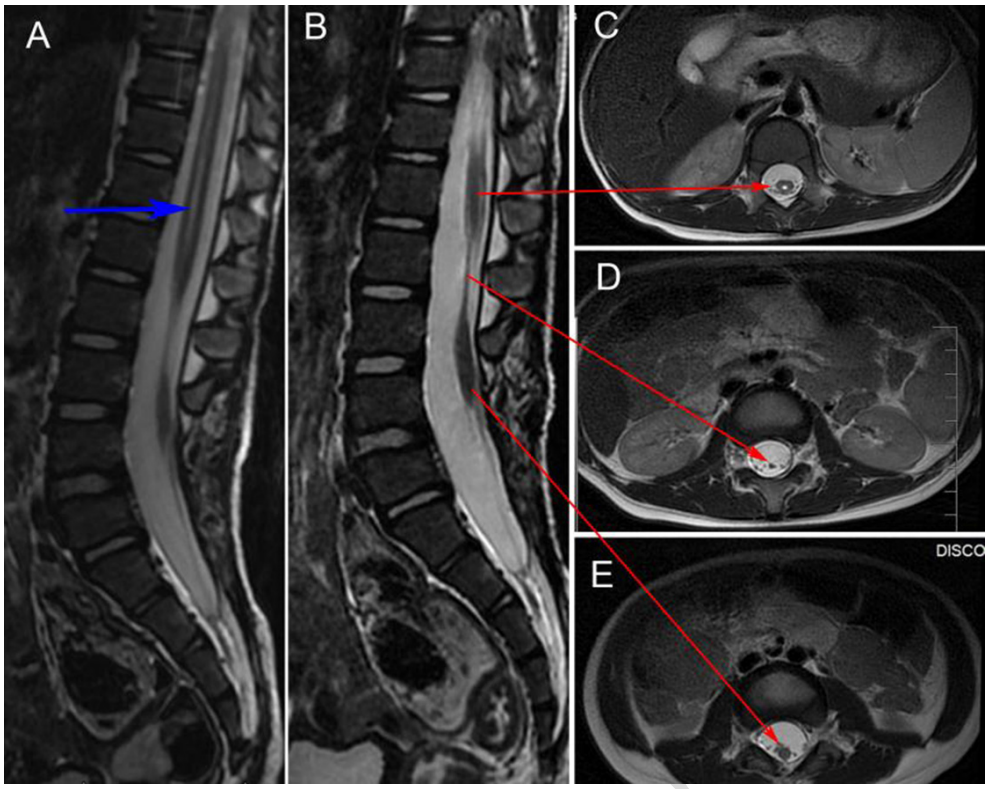

Fig. 3. Postoperative T2-weighted MRI of patient 2. A : Sagittal view shows an enlarged central canal (blue arrow) at the upper spinal cord. B : Sagittal view shows the upper spinal cord (L_2) and the lower spinal cord ($L_{3/4}$) and the fibrous band connecting them. C : Axial view at L_1 shows the upper spinal cord and the enlarged central canal. D : Axial view at L_2 shows the connecting fibrous band between the “upper spinal cord” and the “lower spinal cord.” E : Axial view shows the lower spinal cord.