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CASE REPORT Open Access

Split notochord syndrome presenting as dorsal spinal mass in a neonate: a case report with characteristic MRI findings

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Abstract

Background Split notochord syndrome is an extremely rare and controversial congenital anomaly considered part of the broader spectrum of complex spinal dysraphism and neurenteric developmental disorders. It results from abnormal embryological separation of the endoderm and ectoderm, leading to a range of vertebral, spinal cord, and potential enteric malformations. Due to significant overlap with other dysraphic conditions, diagnosis is primarily radiological and often challenging.

Case presentation We report a case of a 10-day-old female neonate presenting with a dorsal lumbosacral swelling noted since birth, delivered at home to a mother with no antenatal care. Physical examination revealed a midline, skin-covered spinal mass without cerebrospinal fluid leakage. MRI of the spine, performed using a low-field (0.5 Tesla) system, demonstrated a midline vertebral defect, widening and segmentation abnormalities of posterior elements, and duplication of the spinal cord, with additional features suggestive of severe spinal dysraphism within the neurenteric spectrum. The diagnosis was based solely on imaging findings without surgical or histopathological confirmation. The constellation of findings was most consistent with probable split notochord syndrome within the neurenteric spectrum. The neonate received supportive management but deteriorated and died after seven days of hospitalization.

Conclusion This case highlights a severe form of spinal dysraphism within the split notochord/neurenteric spectrum diagnosed on MRI in a resource-limited setting. It underscores the importance of early antenatal surveillance and postnatal imaging for prompt recognition of complex congenital spinal anomalies. MRI remains the key diagnostic modality for defining the extent of disease and guiding management, although overlap between entities within this spectrum may limit strict diagnostic classification.

Keywords Split notochord syndrome, Spinal dysraphism, Neonate, MRI, Congenital anomaly, Case report

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Introduction

As a rare form of spinal dysraphism, split notochord syndrome features a split vertebral column that creates a double spinal column. This malformation stems from a failure of the lateral ossification centers of the vertebral bodies to fuse [1, 2]. The defect is thought to occur during the gastrulation stage in the third week of embryogenesis [3]. The condition represents a spectrum ranging from isolated neurenteric cysts to complex malformations that include vertebral clefts, duplicated spinal cords, and dorsal enteric fistulas [2, 4]. Split notochord syndrome remains an exceptionally rare congenital disorder, with most available evidence derived from isolated case reports and small literature reviews, highlighting the diagnostic challenges and the need for multidisciplinary recognition of these complex developmental anomalies [5]. Magnetic resonance imaging plays a pivotal role in delineating the extent of anomalies, associated dysraphic features, and possible communication between neural and enteric structures, making it the imaging modality of choice [6, 7]. This case report has been prepared in accordance with CARE checklist.

Case presentation

Patient information

A 10-day-old female neonate presented with a swelling over the lower back since birth. The child was delivered at home to a 30-year-old primigravida mother from a rural area with no antenatal care follow-up or prenatal imaging. The birth weight, gestational age, and APGAR scores were not documented due to home delivery. No significant antenatal, perinatal, medical, surgical, or family history was reported.

Clinical findings

Physical examination revealed a midline dorsal swelling in the lumbosacral region that was soft, non-pulsatile, and covered with intact skin. There was no evidence of cerebrospinal fluid leakage. Neurological examination revealed reduced spontaneous movement of both lower limbs with suspected hypotonia. Assessment of bowel and bladder function was not feasible given the neonatal age and clinical condition.

Timeline

The sequence of clinical events from birth to outcome is summarized in Table 1.

Diagnostic assessment

In view of the clinical findings, a conventional MRI of the lumbosacral spine was performed using a low-field (0.5 Tesla) scanner for further anatomical delineation (Figs. 1 and 2). The study included sagittal T1-weighted, sagittal T2-weighted, sagittal short tau inversion recovery (STIR), and axial T2-weighted sequences. Imaging revealed a complex congenital anomaly involving the lumbosacral spine. A midline posterior vertebral defect was identified predominantly involving the lower lumbar to upper sacral levels (L4–S1), characterized by widening and splitting of the posterior spinal elements (Fig. 1A–C). Through this defect, herniation of bowel loops was observed extending beyond the confines of the spinal canal, as demonstrated on sagittal T2-weighted and STIR images (Fig. 1B, C). These findings correlate with the figure legends, which illustrate the osseous defect and exteriorization of intestinal structures.

In addition to the vertebral abnormality, a myelomeningocele was identified at the level of the defect, best appreciated on axial T2-weighted imaging (Fig. 2A). Furthermore, there was duplication of the spinal cord over a short segment, with two hemicords visualized, consistent with split cord malformation (diastematomyelia) (Fig. 2B).

No definite osseous or fibrous septum separating the hemicords was clearly demonstrated on the available sequences.

The conus medullaris appeared low-lying, raising the possibility of an associated tethered cord, although precise characterization was limited by the spatial resolution of the low-field MRI system.

Despite the presence of enteric herniation, a definite neurenteric fistulous tract could not be directly visualized.

Overall, the constellation of imaging findings, including a midline vertebral defect, herniation of bowel loops, myelomeningocele, and spinal cord duplication, is most consistent with a probable diagnosis within the split notochord syndrome spectrum, although definitive confirmation was not possible in the absence of surgical or histopathological correlation.

Diagnostic challenges

The absence of antenatal imaging and limited availability of advanced diagnostic resources posed challenges in early detection and comprehensive evaluation.

Table 1 Timeline of clinical events

Time Point	Clinical Event Description
Birth	Home delivery without antenatal care; dorsal lumbosacral swelling noticed at birth
Day 1–10	Persistent swelling without medical evaluation
Day 10	Presentation to hospital; physical examination performed
Day 10	MRI of the lumbosacral spine performed for diagnostic evaluation
Day 10–17	Hospital admission and supportive management initiated
Day 17	Clinical deterioration and death

Therapeutic intervention

The patient was admitted for supportive management, including thermal care, intravenous fluids, and close monitoring. Empiric antibiotic therapy was considered due to infection risk. Neurosurgical intervention was indicated but was not feasible due to limited resources. Neurosurgical consultation was not available locally, and referral to a higher-level center was considered; however, this was not achieved due to logistical and resource constraints.

Follow-up and outcomes

The patient's condition progressively worsened during hospitalization. Despite supportive care, she died after seven days. The exact cause of death could not be definitively determined. Severe congenital malformation with associated complications likely contributed to the fatal outcome.

Discussion

Split notochord syndrome is an exceptionally rare congenital malformation within the spectrum of complex spinal dysraphism and neurenteric developmental anomalies. The pathogenesis of split notochord syndrome is believed to involve abnormal interactions between the developing notochord, endoderm, and ectoderm during early gastrulation, resulting in variable combinations of vertebral defects, spinal cord abnormalities, and enteric malformations. Contemporary literature supports viewing split notochord syndrome as part of a broader

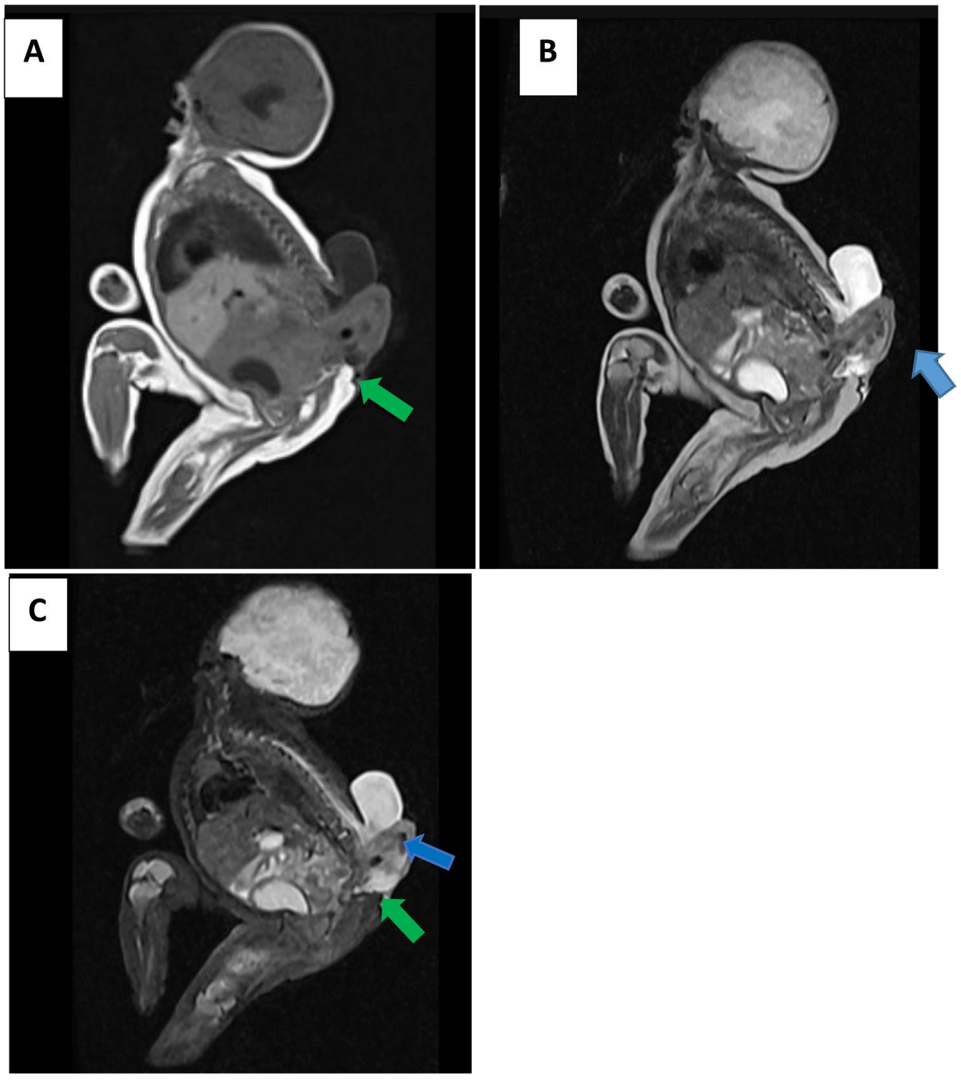


Fig. 1 Sagittal MRI of the lumbosacral spine demonstrating features of split notochord syndrome. **A** Sagittal T1-weighted image showing a posterior midline vertebral defect with widening and splitting of the posterior spinal elements at the lumbosacral region (green arrow) with widening and splitting of the posterior elements. **B** Sagittal T2-weighted image demonstrating herniation of bowel loops through the vertebral defect (blue arrows) with associated spinal dysraphism. **C** Sagittal STIR image confirming the vertebral defect (green arrow) and exteriorized bowel loops (blue arrows)

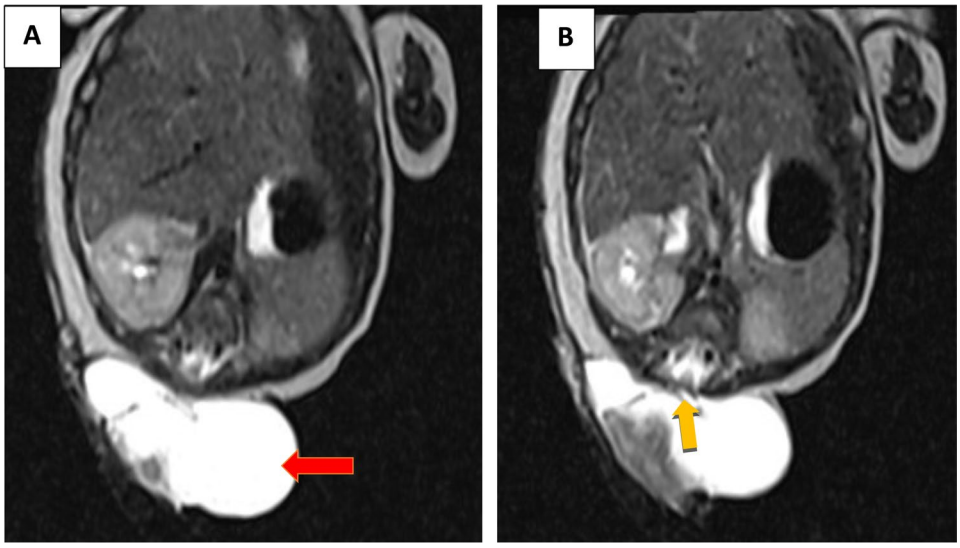


Fig. 2 Axial T2-weighted MRI of the lumbosacral spine demonstrating associated anomalies. **A** Axial image showing a myelomeningocele at the level of the defect (red arrow). **B** Axial image demonstrating splitting of the spinal cord consistent with diastematomyelia, with two hemicords identified (orange arrow)

neurenteric developmental spectrum rather than an isolated malformation [4, 5, 8]. This embryological defect leads to a wide range of anomalies involving the vertebral column, spinal cord, and gastrointestinal tract, with clinical severity depending on the degree of developmental disruption [2–4].

In the present case, MRI demonstrates findings that strongly support a diagnosis within the split notochord syndrome spectrum rather than a definitive diagnosis. These includes a midline vertebral defect with widening and splitting of the posterior elements, herniation of intestinal loops through the spinal defect, a coexisting myelomeningocele, and duplication of the spinal cord consistent with diastematomyelia. The simultaneous presence of neural and enteric components is particularly significant, as it reflects the hallmark feature of split notochord syndrome, which is a persistent communication between the developing gastrointestinal tract and neural structures, although direct communication could not be conclusively demonstrated in this case [3, 8]. The identification of exteriorized intestinal loops through the vertebral defect represents a severe manifestation and places this case at the more complex end of the neurenteric spectrum [4, 7].

Although no surgical or histopathological confirmation is obtained, the diagnosis in this case is established radiologically based on characteristic imaging findings. MRI plays a central role in such cases, although its ability to demonstrate subtle neurenteric fistulous communication may be limited, particularly in low-field systems. It allows detailed evaluation of spinal cord morphology, vertebral anomalies, and associated soft tissue components, including potential enteric involvement [6, 7]. In fact, several reports emphasize that split notochord syndrome is frequently diagnosed on imaging alone, particularly in settings where surgical correlation is not feasible [7]. Nevertheless, due to the considerable overlap between split notochord syndrome and other neurenteric-associated malformations, the diagnosis should be interpreted within the broader spectrum of notochordal developmental disorders [2, 4, 8].

A comparison with previously reported neonatal cases of split notochord syndrome and related neurenteric-spectrum malformations is summarized in Table 2. Prior reports have described varying combinations of vertebral defects, spinal cord duplication, and neurenteric fistulas, with most cases undergoing surgical intervention. For example, cases reported by Srivastava et al. demonstrated clear neurenteric fistulous communication and were managed surgically with favorable outcomes. In contrast, other reports such as Wakao et al. and Alelyani et al. described spinal cord duplication and associated anomalies without overt enteric fistulization. Compared to these cases, the present case represents a more severe phenotype characterized by external herniation of bowel loops through a vertebral defect, a feature less commonly documented. Furthermore, unlike most previously reported cases, surgical intervention was not feasible in our setting, which likely contributed to the unfavorable outcome.

The differential diagnosis in this case includes split cord malformation (diastematomyelia), isolated myelomeningocele with associated vertebral defects, neurenteric cysts, and caudal duplication syndrome. Recent reviews of caudal duplication syndrome emphasize that this entity encompasses a spectrum of hindgut, genitourinary,

Table 2 Comparison of reported neonatal cases of split notochord syndrome / neurenteric spectrum malformations

Author (Year)	Age at Presentation	Key Imaging Findings	Enteric Involvement	Associated Anomalies	Management	Outcome
Srivastava et al. (2010) [2]	Neonate	Vertebral defect with neuroenteric fistula	Yes	Myelomeningocele	Surgery	Improved
Wakao et al. (2011) [7]	Infant	Split cord, vertebral anomalies	No clear fistula	Diastematomyelia	Surgery	Stable
Jasiewicz et al. (2020) [4]	Neonate	Spine duplication, complex dysraphism	Variable	Vertebral duplication	Surgery	Variable
Alelyani et al. (2022) [3]	Neonate	Spinal column duplication, lipoma	No	Lipoma, duplication anomalies	Surgery	Survived
Present Case (2026)	10-day-old neonate	Midline vertebral defect (L4–S1), spinal cord duplication, bowel herniation	Yes (externalized bowel loops, no definite fistula)	Myelomeningocele, probable tethered cord	Supportive only (no surgery)	Died

Key Comparative Insights

- The present case represents a more severe phenotype, with external bowel herniation, which is less commonly reported
- Unlike most reported cases, no surgical intervention was feasible, significantly impacting outcome
- Many published cases include confirmed neurenteric fistulas, whereas in this case, direct communication could not be demonstrated radiologically
- The coexistence of vertebral defect, spinal cord duplication, and enteric involvement aligns with the severe end of the neurenteric spectrum

vertebral, and spinal cord duplications, which may overlap clinically with other neurenteric developmental disorders [2, 5, 8]. Although duplication of the spinal cord was observed, the absence of a clearly defined osseous or fibrous septum separating two hemicords, together with the presence of enteric herniation, makes isolated split cord malformation less likely. The classification of split cord malformation (SCM) into two distinct phenotypes hinges on the structural division of the dural sac and the histological makeup of the midline septum. While Type I SCM presents with a duplicated dural sac compartmentalized by an osseous septum, Type II variants are defined by a singular dural sac anchored by a fibrous band. Recognizing this anatomical divergence is critical when discussing surgical approaches and patient outcomes [9]. Similarly, while a myelomeningocele was identified, isolated cases do not typically demonstrate herniation of intestinal loops through a vertebral defect, which is a key distinguishing feature in this case. Neurenteric cysts generally present as cystic intradural lesions without external bowel herniation or complex vertebral defects, further differentiating them from the current findings. Caudal duplication syndrome, on the other hand, is characterized by duplication of distal gastrointestinal and genitourinary structures, which were not identified clinically or radiologically in this patient [5]. Taken together, the coexistence of vertebral defect, spinal cord duplication, and enteric involvement strongly favors a diagnosis within the split notochord syndrome spectrum [3, 4, 8].

Magnetic resonance imaging remains the gold standard for evaluating complex spinal dysraphism due to its superior soft tissue resolution and multiplanar capability [6, 7]. Importantly, in this case, imaging performed on a low-field (0.5 Tesla) MRI system was sufficient to identify the key diagnostic features, highlighting its utility in resource-limited settings. However, low-field MRI may have limitations in spatial resolution, and subtle findings such as small neurenteric fistulous tracts or intradural anomalies may not be fully delineated [6]. Additional imaging with higher-field MRI or complementary computed tomography for osseous detail could further enhance anatomical characterization when available [7].

Management of split notochord syndrome is complex and typically requires early multidisciplinary intervention, including neurosurgical repair of spinal defects, management of associated enteric abnormalities, and prevention of complications such as infection and progressive neurological deterioration [2, 10]. Early diagnosis is crucial, as outcomes are significantly improved when surgical intervention is performed before the onset of secondary complications [10]. In the present case, delayed presentation, absence of antenatal care, and limited access to specialized surgical management contributed to a poor outcome. The patient deteriorated rapidly and died within seven days of hospitalization. Although the exact cause of death could not be definitively established, it was likely multifactorial, including a severe congenital malformation, risk of infection, and progressive neurological compromise.

This case highlights several important clinical and academic considerations. First, it demonstrates a severe form of split notochord syndrome with classical MRI features, including combined vertebral, neural, and enteric abnormalities. Second, it emphasizes the diagnostic value of MRI even when performed with limited resources. Third, it underscores the critical importance of antenatal care and early postnatal imaging in detecting life-threatening congenital anomalies [6, 7, 11]. Finally, given the rarity of split notochord syndrome and ongoing debate regarding its classification, this case supports the concept that split notochord syndrome

represents a spectrum of neurenteric developmental disorders rather than a single discrete entity [2–5].

Patient perspective

The patient's guardian reported significant distress due to the child's condition and lack of access to specialized care. The family expressed concern over the absence of antenatal evaluation and consented to publication to improve awareness of similar conditions.

Strengths and limitations

This case has several limitations. First, the diagnosis was based solely on imaging findings without surgical or histopathological confirmation, which may limit definitive classification within the neurenteric spectrum. Second, the use of a low-field (0.5 Tesla) MRI system may have restricted the detection of subtle anatomical details. Third, limited clinical data and absence of long-term follow-up further constrain comprehensive evaluation. Additionally, the absence of both surgical intervention and autopsy further limits definitive diagnostic confirmation. Despite these limitations, the case provides valuable insight into a rare and severe presentation of split notochord syndrome. It also highlights the diagnostic utility of MRI and underscores the importance of early recognition of complex spinal dysraphism.

Conclusion

Split notochord syndrome is a rare and severe congenital anomaly within the spectrum of complex spinal dysraphism, characterized by the coexistence of vertebral defects, spinal cord malformations, and enteric involvement. The present case demonstrates a radiologic presentation suggestive of split notochord syndrome, including a midline vertebral defect, herniation of intestinal loops, myelomeningocele, and diastematomyelia. MRI remains the cornerstone of diagnosis, particularly when surgical confirmation is not feasible, providing detailed anatomical characterization essential for clinical decision-making. Early detection through antenatal screening and timely postnatal imaging is critical, as prompt surgical intervention may significantly improve outcomes. The fatal outcome in this case underscores the serious consequences of delayed diagnosis and limited access to specialized care. Increased awareness of split notochord syndrome and its characteristic imaging features is essential for early recognition, appropriate classification within the neurenteric spectrum, and improved clinical outcomes.

Supplementary Information

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Supplementary Material 1.

Institutional review board statement

The study did not require ethical approval.

Authors' contributions

Conceptualization, data curation, project administration: BMB, figure preparation: SAB; Writing original draft, writing review and editing: BMB and SAB.

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Data availability

No datasets were generated or analysed during the current study.

Declarations

Consent for publication

Written informed consent has been obtained from the guardian to publish this paper.

Competing interests

The authors declare no competing interests.

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