



Residência **RP** Pediátrica

Publicação Oficial da Sociedade Brasileira de Pediatria

ISSN-Online: 2236-6814

Submitted on: 03/17/2025

Approved on: 12/02/2025

CASE REPORT

Secondary enuresis and chronic kidney disease in adolescent with anchored spinel cord: case report

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Keywords:

Spinal dysraphism,
Neural Tube Defects,
Renal Insufficiency,
chronic,
Enuresis.

Abstract

Objective: To describe a clinical case of an adolescent with a late diagnosis of occult spinal dysraphism, highlighting the clinical manifestations that led to suspicion, confirmation of the diagnosis, and complications. **Case report:** Adolescent with a history of enuresis and recurrent urinary infections that began at 10 years of age. She denied previous urinary symptoms. At 11 years of age, she was hospitalized due to pyelonephritis and renal failure. Physical examination revealed deviation of the intergluteal sulcus and claw toes. Ultrasound of the kidneys and urinary tract showed ureterohydronephrosis, voiding cystourethrography showed bilateral vesicoureteral reflux grade V, urodynamic evaluation showed detrusor overactivity and vesicoesphincter dyssynergia, and magnetic resonance imaging of the spine showed lipomyelomeningocele. **Comments:** Occult dysraphism is a congenital malformation of the spinal cord covered by intact skin. In tethered spinal cord, there is loss of mobility of the spinal cord and, as it grows, it stretches and the appearance of urological, intestinal and lower limb symptoms, characterizing tethered spinal cord syndrome. Diagnosis of tethered spinal cord early in life is essential for good evolution and prevention of complications that are often irreversible. However, at this stage, diagnosis can be challenging, and it is necessary to be aware of the presence of neurocutaneous stigmata in the lumbosacral region, symmetry of the intergluteal groove and lower limbs.

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INTRODUCTION

Neural tube defects (NTDs) are the most common congenital malformations after congenital heart diseases. Occult spinal dysraphism (OSD) is characterized by failure of posterior vertebral fusion with intact overlying skin, differing from open dysraphisms, such as myelomeningocele, in which neural tissue is exposed. Although the skin covers the malformation, cutaneous changes may indicate the diagnosis and are known as neurocutaneous stigmata. The presence of these cutaneous markers is due to the fact that the central nervous system and the ectoderm share the same embryological origin.

Occult dysraphisms may be associated with abnormal fixation of part of the spinal cord—usually the conus medullaris—to structures within the vertebral canal (tethered cord). Tethered cord syndrome is a constellation of neurological, orthopedic, urological, and bowel symptoms secondary to stretching of the spinal cord with growth, leading to reduced blood flow to the conus medullaris and nerve injury.

The pubertal growth spurt, due to its high growth velocity, leads to spinal cord stretching, nerve injury, and worsening of signs and symptoms, often irreversibly. Early diagnosis, ideally during infancy, allows intervention before significant cord stretching occurs. However, at this stage, the only signs may be cutaneous changes in the lumbosacral region or deviation of the intergluteal cleft, making OSD a diagnostic challenge.

In this article, we describe the clinical case of a patient with signs suggestive of occult spinal dysraphism and late diagnosis in adolescence, leading to irreversible urinary tract complications, with the aim of alerting pediatricians to the signs and symptoms that warrant early diagnostic investigation.

The study was approved by the institution's Research Ethics Committee on 08/28/2024 under number 82533524.3.0000.5259.

CASE DESCRIPTION

Patient FSSO, female, 11 years old, previously healthy, was first evaluated due to secondary enuresis with progressively increasing frequency until it became daily, and recurrent urinary tract infections beginning at 10 years of age, with no prior history. She also reports increased urinary frequency, a sensation of incomplete bladder emptying, urinary urgency, episodes of urinary incontinence, and constipation. Pregnancy, delivery, and developmental history were unremarkable. She denies a family history of enuresis.

On physical examination, she was eutrophic (BMI Z-score between 0 and -1), with weight (Z-score between 0 and -1) and height (Z-score 0) appropriate for age, blood pressure of 110×70 mmHg, and pubertal development at Tanner stage M2P3. Deviation of the intergluteal cleft (Figure 1), a

palpable bladder, and claw toes (Figure 2) were observed. On neurological examination, reflexes, tone, and muscle strength were preserved, with normal gait and balance.

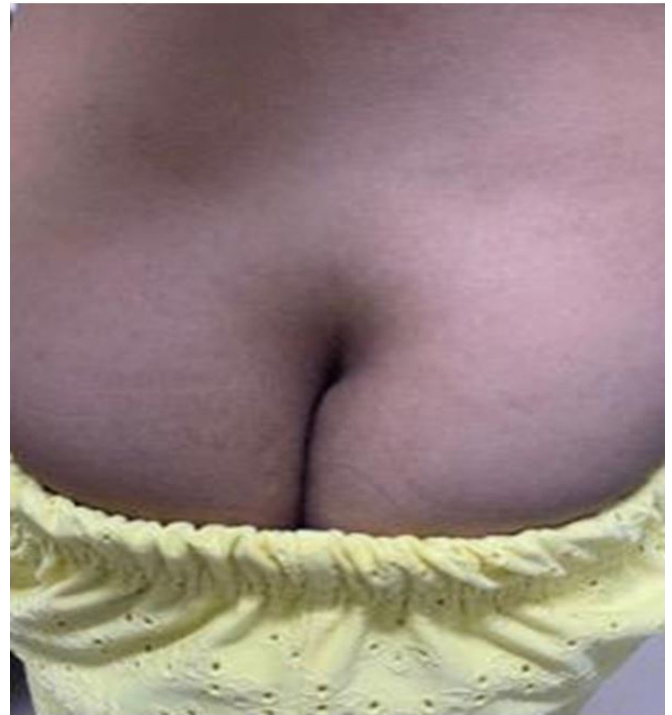


Figure 1. Deviation of the intergluteal cleft.



Figure 2. Claw toes.

At admission, the patient had laboratory findings showing mild anemia, leukocytosis, elevated C-reactive protein, urea of 143 mg/dL, and creatinine of 3.6 mg/dL, with a creatinine clearance of 19 mL/min/1.74 m² (G4).

Ultrasound performed during hospitalization showed dilatation of the pelvicalyceal system and ureter, a distended bladder with thickened and trabeculated walls, and diverticular images.

The working hypotheses were tethered cord syndrome, high-pressure neurogenic bladder secondary to spinal cord stretching, and chronic kidney disease. The following complementary tests were performed:

- Renal and urinary tract ultrasound: ureterohydronephrosis, enlarged kidneys, a distended bladder with trabeculated walls, and increased post-void residual for age.

- Voiding cystourethrography: bilateral grade V vesicoureteral reflux (VUR) and a trabeculated bladder (Figure 3).

- Lumbosacral spine magnetic resonance imaging: The spinal cord is tethered at the S1/S2 level and is associated with an expansile lesion with high signal intensity on T1- and T2-weighted images. The most nodular component measures approximately 2.6 × 2.3 × 1.5 cm, also at the S1/S2 level, with signal loss on fat-saturation sequences and no post-contrast enhancement. There is possible inferior and leftward extension of the fatty lesion to S3/S4. These findings are suggestive of a lipoma. Incomplete closure of the lower sacrum is noted. The spinal canal is widened in the sacral region. Findings are consistent with lipomyelocoele (Figure 4).



Figure 3. Voiding cystourethrography.

Caption: Bilateral grade V VUR and a trabeculated bladder wall are observed.



Figure 4. Magnetic resonance imaging.

Caption: Presence of a lipoma and spinal cord tethering.

- Urodynamic study: detrusor overactivity and detrusor-sphincter dyssynergia.

- The diagnosis of tethered cord syndrome, high-pressure neurogenic bladder (NB), and chronic kidney disease was established. NB was determined to be secondary to occult dysraphism and spinal cord stretching, whereas bilateral grade V VUR and kidney disease were complications resulting from NB. Clean intermittent catheterization, nocturnal bladder drainage, and anticholinergic therapy were initiated, resulting in a reduction in urinary tract infection episodes and partial improvement in renal function, with creatinine of 1.1 mg/dL and a creatinine clearance of 79 mL/min/1.74 m² (G2). The patient was referred to Pediatric Neurosurgery and underwent surgery for resection of the conus medullaris.

COMMENTS

Tethered cord syndrome (TCS) may result from open dysraphisms or from occult spinal dysraphism (OSD), as in the case described. Excessive traction on the distal spinal cord compromises mitochondrial oxidative metabolism, causing hypoxia and worsening dysfunction as traction increases, thereby worsening prognosis^{1,2}.

Lipomyelomeningocele, identified on magnetic resonance imaging, is a fat-containing mass in the intervertebral space that prevents the normal upward movement of the conus medullaris during childhood development and may result in neurological sequelae involving the urinary tract and lower limbs. Although this occult dysraphism is rare, it should be considered in children with an abnormal voiding pattern,

recently onset urinary incontinence—especially during puberty—and recurrent urinary tract infections^{2,3}.

Symptoms of TCS vary according to age group and may involve multiple systems, most commonly cutaneous, orthopedic, neurological, anorectal, and urological systems. The adolescent growth spurt is associated with worsening and/or onset of symptoms and often irreversible complications^{1,2}.

In infants, cutaneous markers (dimples or pits, lipomas, asymmetry of the intergluteal cleft, and hair tufts) are the most common findings. Approximately 80% of newborns with OSD present with a sign in the lumbosacral region, whereas only 3% of healthy newborns will have classic lesions. These may be the only clinical signs and should be assessed during routine physical examination. Neurological and orthopedic findings are uncommon at this stage^{1,4–6}.

In preschool children, difficulty with sphincter control may be a warning sign. In school-age children and adolescents, gait changes and foot deformities (pes cavus and claw toes/hammer toes), back pain, limited trunk flexion, and progression of scoliosis may be noted, as well as urinary incontinence, enuresis, recurrent urinary tract infections, constipation, and/or fecal incontinence^{1,2,4,6}.

With growth, urological symptoms become more evident. However, lower urinary tract dysfunction (LUTD) may be present at any age and can be detected by urodynamic evaluation^{1,2,4,6}.

Urodynamic evaluation is the gold standard for urological assessment in patients with OSD and should be performed in all suspected cases of TCS. This examination allows detection of subclinical dysfunction, assessment of severity, and guidance of treatment⁷.

Neurogenic bowel dysfunction (neurogenic bowel) is often associated with LUTD (neurogenic bladder) and manifests as constipation and fecal incontinence^{1,2,4,6}.

Neurosensory symptoms may occur, such as weakness in the feet or legs, with an approximate frequency of 12%. Hypo- or hyperreflexia, muscle atrophy, spasticity, decreased proprioception, and painless ulcers have also been described. Delayed or abnormal gait may occur, particularly asymmetrically^{1,8}.

Orthopedic abnormalities are common in TCS, affecting approximately 90% of these patients. Scoliosis is the most common (25%), followed by limb-length discrepancy (6%) and asymmetry or deformity of the feet (6%), as in the case described, with pes cavus and claw/hammer toes¹.

The imaging modality of choice for investigating suspected OSD is magnetic resonance imaging, ideally of the neuroaxis (brain and spine)¹. Lumbosacral ultrasonography may be used as a screening tool in the first three months of life, before ossification of the posterior sacral elements¹.

The degree of spinal cord traction is directly related to prognosis and may lead to irreversible lesions; therefore,

some studies indicate that untethering should be performed as soon as the diagnosis is established to prevent permanent injury^{1,9–11}.

A highly important component of neurogenic bladder management is clean intermittent catheterization (CIC). When indicated, it should be initiated as early as possible. CIC reduces the risk of renal complications by allowing bladder emptying and reducing symptomatic urinary tract infections⁷. Pharmacological treatment with antimuscarinics should be used to control detrusor overactivity and intravesical pressure⁷.

CONCLUSION

Occult spinal dysraphism (OSD) and tethered cord syndrome (TCS) present a diagnostic challenge. Pediatricians must be attentive to indicators in the medical history and physical examination, as delayed diagnosis can lead to irreversible sequelae, as in the case described. Attention should begin at birth by identifying cutaneous stigmata suggestive of OSD and should continue throughout growth, investigating urological, orthopedic, neurological, and anorectal abnormalities, particularly during the pubertal growth spurt, the period of highest risk for TCS manifestation. Early diagnosis and treatment could prevent chronic kidney disease, a condition that is irreversible once established.

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