Spontaneous improvement in urological dysfunction in children with congenital spinal lipomas of the conus medullaris

Report of 2 cases

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Congenital spinal lipomas of the conus (SLCs) are among the most common closed neural tube defects. The treatment of SLC is an area of controversy because the true natural history of this condition is unknown. Here, the authors present two cases of SLC presenting in infancy with compromised lower urinary tract function, which was objectively confirmed by abnormal urodynamic studies. In both cases, there was spontaneous improvement in urodynamic parameters, with stable normal urinary function at the long-term follow-up. Although cases of spontaneous radiological regression of SLC have very infrequently been reported, they have not been associated with the reversal of already present neurological deficits. This report reinforces the need for further delineation of the true natural history of SLC and highlights the dynamic nature of associated neurological compromise over time. (http://thejns.org/doi/abs/10.3171/2014.2.PEDS13519)

KEY WORDS • spinal dysraphism • spinal lipoma • tethered cord • urinary dysfunction • urodynamics • spine • congenital

Congenital spinal lipomas of the conus (SLCs) are among the most common closed neural tube defects. The treatment of SLC is an area of controversy because the true natural history of this condition is unknown. Here, the authors present two cases of SLC presenting in infancy with compromised lower urinary tract function, which was objectively confirmed by abnormal urodynamic studies. In both cases, there was spontaneous improvement in urodynamic parameters, with stable normal urinary function at the long-term follow-up. Although cases of spontaneous radiological regression of SLC have very infrequently been reported, they have not been associated with the reversal of already present neurological deficits. This report reinforces the need for further delineation of the true natural history of SLC and highlights the dynamic nature of associated neurological compromise over time.

 Abbreviations used in this paper: CIC = clean intermittent catheterization; SLC = spinal lipoma of the conus; UDS = urodynamics study.

Case Reports

Case 1

History and Examination. This male infant presented shortly after birth with a subcutaneous mass and two small overlying cutaneous hemangiomas in the lower lumbar region. The infant appeared to be urinating and defecating without any issues. Neurological examination, including anal tone, was unremarkable apart from an absent ankle jerk reflex in the left leg. Magnetic resonance imaging of the spine demonstrated a low-lying conus medullaris at the L-5 level. A large intraspinal lipoma was
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seen extending from the L-3 to S-4 levels and communicating with a fatty subcutaneous collection via dysraphic sacral vertebrae (Fig. 1).

Urology Assessment. A urodynamics study (UDS) was performed when the boy was 6 months old. He had elevated postvoid residual volumes of 45–50 ml (reference < 5 ml). In addition, he voided with high pressures in the range of 100 cm H\textsubscript{2}O (reference 50–70 cm H\textsubscript{2}O). Electromyography revealed incomplete sphincter relaxation. There was also evidence of detrusor overactivity on cystometry. A voiding cystourethrogram demonstrated a dilated posterior urethra; thus, cystoscopy was performed, which ruled out the presence of posterior urethral valves. Serial renal and bladder ultrasounds were unremarkable, without evidence of hydronephrosis or parenchymal abnormality. Given the above evidence of lower urinary tract neurogenic dysfunction, oxybutynin was commenced for high voiding pressures and detrusor instability, and clean intermittent catheterization (CIC) was performed for incomplete bladder emptying.

Clinical Course. Surgical intervention was discussed with the patient’s parents, but nonoperative management with close clinical follow-up was ultimately chosen. By 11 months of age, the boy’s postvoid residual volumes had decreased to approximately 10 ml; therefore, CIC was discontinued. Oxybutynin was stopped soon thereafter, and he has since been urinating spontaneously without any trouble. A repeat UDS when he was 4 years old showed no evidence of urinary dysfunction, and he was completely toilet trained. Follow-up spine MRI performed when the boy was 7 years of age demonstrated a reduction in the size of the intraspinal lipoma, with a persistent low-lying conus medullaris at the L-5 spinal level (Fig. 2). At the last follow-up, this child continued to have normal lower limb strength and sensation, without evidence of bladder or bowel dysfunction after 9 years of serial observation.

Case 2

History and Examination. This male infant presented shortly after birth with a skin-covered lumbosacral mass. The child’s neurological examination was entirely normal, including lower limb function. Spinal MRI revealed the tip of the conus medullaris was at the level of the L-4 vertebral body. A substantial amount of fat extended from the intradural space of the lumbar and sacral spinal canal, through a posterior 4-mm bony defect at the L5–S1 spinal level, and into a subcutaneous lipomatous collection, consistent with a diagnosis of SLC (Fig. 3).

Urology Assessment. When the boy was 2 months of age, a voiding cystourethrogram demonstrated left-sided Grade II vesicoureteral reflux, with retrograde flow of urine into the renal pelvis and calyces. Cystometry revealed uninhibited high-amplitude bladder contractures during filling, with eventual urinary leak through the urethra. Renal and bladder ultrasounds were unremarkable. Additionally, the parents reported frequent urination with small output volumes. Consequently, we started CIC, oxybutynin for detrusor hyperreflexia, and prophylactic antibiotics for vesicoureteral reflux.

Clinical Course. Surgical repair of the spinal lipoma was offered on the basis of the clinical signs and abnormal UDS. However, the parents opted for nonoperative treatment. Thus, the boy was managed with serial clinical observation. A UDS at 2 years of age showed persistent overactive bladder with substantial instability, and thus the boy remained on oxybutynin and prophylactic antibiotics. At 4 years of age, he was completely toilet trained and did not have any urinary frequency, urgency, incontinence, or infections. A follow-up evaluation revealed nor-
mal urinary function, with a peak flow rate of 8.8 ml/sec-
ond, volume voided of 129 ml, and no signs of detrusor
instability or detrusor-sphincter dyssynergia. Oxybutynin
and antibiotics were eventually discontinued. Follow-up
flow-rate studies when the boy was 6 and 8 years old
again revealed normal bladder emptying. Routine spinal
MRI performed at 10 years of age revealed a persistent
low-lying conus at the L-4 level and an interval increase
in the size of the known lipoma (Fig. 4). At his 10-year
follow-up, the child remained neurologically intact, with
no symptoms of urinary dysfunction.

Discussion

The decision of whether and when to surgically treat
SLC remains a topic of heated debate. Several au-
thors have advocated for prophylactic cord detethering
surgery in asymptomatic patients. One purported rationale for this relates to evidence suggest-
ing that once neurological deficits have manifested, only
a minority of these patients will improve after surgery,
whereas asymptomatic patients only rarely develop symp-
toms after prophylactic surgery. There is, however, a
paucity of data pertaining to the natural history of SLC
and tethered cord. To date, there are only two published
series of nonoperatively managed SLC—53 cases from
Necker-Enfants Malades Hospital, Paris, and another
56 cases from Great Ormond Street Hospital, London.
Both of these natural history studies indicate a more be-
nign clinical course for SLC than previously believed.

Neurogenic lower urinary tract dysfunction is con-
sidered one of the most frequent manifestations of con-
genital SLC. Associated clinical symptoms can include
urinary incontinence, urinary tract infections, and/or
urinary retention. However, as illustrated by the two
cases featured here, children with spinal dysraphism may
remain essentially asymptomatic despite the presence of
significant urological dysfunction, which speaks to the
value, and arguably the necessity, of UDS in evaluating
these patients. Urologists use UDS to detect lower urinary tract dysfunction so that interventions (for exam-
ple, prophylactic antibiotics, CIC, anticholinergics, and
so forth) can be implemented to preserve the integrity of
the upper urinary tract (that is, the kidneys). The most
important limitation of pediatric UDS occurs when there
is a lack of cooperation from the child (for example, ir-
ritability, crying, and so forth), which can interfere with
abdominal and vesical pressure monitoring. Even then,
the reported sensitivity, specificity, positive predictive
value, and negative predictive value of UDS in predicting
upper urinary tract deterioration in infants with spina bi-
fida can reach 87%, 93%, 85%, and 81%, respectively.
The most common urodynamic findings in the context of
congenital SLC include detrusor overactivity, detrusor-
sphincter dyssynergia, reduced bladder compliance, and
incomplete emptying.

The prognosis of urological dysfunction in spinal
dysraphic states is poor in older children and adults, in
whom surgery only rarely leads to improvement in symp-
toms. Satar et al. reported no improvement or worsen-
ing of UDS findings in 16 of 19 older patients (mean age
14.5 years) following surgical repair of an occult spinal
dysraphism. By contrast, urological outcomes are more
favorable in infants, with reversal of bladder deficits not
uncommon following cord detethering surgery. Sathi
et al. found normalization of an abnormal preoperative
UDS in 83% of infants following operative repair of SLC.
Similarly, Atala et al. reported postoperative improve-
ment in lower urinary tract function in 9 of 11 infants
with an abnormal preoperative UDS. In any case, the

Fig. 3. Case 2. Sagittal T1-weighted MR images obtained in a
5-month-old boy, demonstrating a spinal lipoma with a low-lying conus
medullaris at the L-4 spinal level.

Fig. 4. Case 2. Sagittal T1-weighted MR images obtained when the
boy was 10 years of age, revealing an interval increase in the size of
the spinal lipoma and a persistent low-lying conus medullaris at the L-4
spinal level.
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presence of bladder and/or bowel deficits often prompts neurosurgical intervention.23,29 For this reason, the natural history of urinary dysfunction in the context of SLC has been difficult to ascertain.

Here, we report on two male infants with objective evidence of urinary dysfunction, as indicated by abnormal UDS. Both children experienced improvement on long-term close monitoring, with evidence of acceptable bladder dynamics without surgical intervention. These patients had no issues or delays in toilet training and have been clinically followed for 9 and 10 years thus far, while remaining free of lower and upper urinary tract disturbance and symptoms. Although a few cases of spontaneous radiological regression of SLC have been reported,8,17,20 to our knowledge, spontaneous improvement of abnormal UDS and urinary dysfunction in congenital SLC has not. Based on our observations, it is very possible that the improvement in urinary function associated with early resection of SLC in infancy may actually reflect the natural history of urological dysfunction in this patient population. Our findings highlight the value of serial urological and neurosurgical assessment of children with this condition, as well as the possible role of conservative management as an option in the absence of progressive neurological deficits or upper urinary tract deterioration.

Conclusions

Our two cases illustrate the possibility of spontaneous reversal of urological dysfunction associated with congenital SLC. These cases reinforce the need for further clarification of the true natural history of SLC.

Disclosure

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

Author contributions to the study and manuscript preparation include the following. Conception and design: Kulkarni, Badihivala, Thompson. Acquisition of data: Kulkarni, Badihivala. Analysis and interpretation of data: all authors. Drafting the article: Badihivala, Thompson. Critically revising the article: all authors. Reviewed submitted version of manuscript: all authors. Approved the final version of the manuscript on behalf of all authors: Kulkarni. Administrative/technical/material support: Kulkarni. Study supervision: Kulkarni.

References

studies in infants less than 1 year old with congenital spinal dysraphism. J Urol 148:584–587, 1992

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