Lumbar lipomyelomeningocele and sacrococcygeal teratoma in siblings: support for an alternative theory of spinal teratoma formation

Report of 2 cases

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Sacrococcygeal teratomas may arise in association with regional developmental errors affecting the caudal embryonic segments and may originate within lumbosacral lipomas. It is therefore possible that sacrococcygeal teratomas and lumbosacral lipomas represent related disorders of embryogenesis. Accordingly, the authors report the cases of 2 siblings. The first child (female) was born with a mature Altman Type III sacrococcygeal teratoma that was resected when she was a neonate. Subsequently, a younger brother was found soon after birth to have an L-4-level lipomyelomeningocele and underwent partial resection and spinal cord untethering at 4 months of age. Although familial forms of each of these conditions have been reported, this is, to the authors’ knowledge, the first reported occurrence of lipomyelomeningocele and sacrococcygeal teratoma in siblings. They propose that an inherited regional tendency to developmental error affecting the caudal embryonic segments was shared by these siblings and resulted in spinal teratoma formation in one of them. (DOI: 10.3171/2010.2.PEDS09502)

Key Words • lipomyelomeningocele • lipoma • sacrococcygeal teratoma • embryogenesis

Case Reports

Case 1

History and Examination. This 2-day-old girl was delivered via caesarian section to a 23-year-old gravida 3, para 0 mother after 36 and 0/7 weeks’ gestation that was complicated by polyhydramnios and diagnosis of a presacral mass on prenatal ultrasound. At birth, the girl was noted to have an irregular 5–6-cm mass palpable at the base of the sacrum. Lower extremity motor strength was symmetrical and full in all groups. Anal sphincter tone was normal; however, she exhibited large postvoid residual volumes necessitating intermittent urinary catheterization. She had no other congenital anomalies except for a possible septate versus bicornuate uterus, which was identified incidentally during subsequent imaging.

Diagnostic Evaluation. The patient’s serum alpha-
fetoprotein level at birth was 163,379 ng/ml. Magnetic resonance imaging of the pelvis with contrast medium revealed a 7.5 × 6.5 × 4.6-cm presacral predominantly cystic mass extending from the L5–S1 level down through the osseous pelvis into the soft tissues of the buttocks, which exhibited peripheral enhancement and scattered thin septations (Fig. 1A and B). There was anterior and superior displacement of the bladder, vagina, uterus, and rectum secondary to the mass.

Surgical Treatment. On Day 2 of the patient’s life, pediatric general surgeons performed surgery for resection of a suspected sacrococcygeal teratoma. After separating the tumor from the overlying subcutaneous tissue and muscle, circumferential dissection was performed to free it from the pelvis. The tumor was centered at the sacrococcygeal joint, necessitating a coccygectomy. The tumor was adherent to the rectum; there was no involvement of the thecal sac. The tumor was completely removed and the pelvic wall reconstructed.

Pathological Examination. The excised mass consisted of irregular, pink-tan, partially membranous tissue with a large cystic area that contained clear fluid and soft yellow-white tissue. The final pathological diagnosis was mature sacrococcygeal teratoma.

Outcome. The patient remained disease free with normal levels of the tumor markers human chorionic gonadotropin and alpha-fetoprotein at 16-month follow-up.

Case 2

History and Examination. Thirteen months later, the only sibling of the patient described in Case 1 was delivered during the 36th week of gestation by repeat cesarean section after an uncomplicated pregnancy. At birth, he was noted to have a 5-mm indented area of waxy, atretic skin with a ring of tiny dysplastic hairs in the midlumbar region that was associated with a small hemangioma. There was no sinus tract or open dysraphism. Findings upon neurological examination were normal and symmetrical throughout, including bilateral lower extremity motor strength, deep tendon reflexes, and sensation. There was no concern for bowel or bladder dysfunction. No other congenital anomalies were identified. At age 4 months, he was referred for MR imaging and neurosurgical consultation.

Radiological Findings. Magnetic resonance imaging of the spine demonstrated a low-lying conus medullaris terminating at the lower half of the L-4 vertebral body (Fig. 1C and D). Additionally, an intradural fat-signal mass consistent with a terminal-dorsal lipomyelomeningocele was identified along the posterior aspect of the distal conus, with extension to the dural surface at L5–S1 and a subcutaneous tract extending to the skin surface overlying L-4. No syrinx or Chiari malformation was observed.

Surgical Treatment. The patient underwent surgery to release the tethered spinal cord and subtotally resect the lipomyelomeningocele, using intraoperative long tract monitoring. After laminoplasty of osseous segments over the cranial end of the intradural lipoma, the dura was opened to the point where it was transversed by the lipoma’s dorsal extension. Adhesions between the dura and lipoma were carefully divided and the transdural extension of the lipoma was transected, effectively untethering the spinal cord, which was noted to relax freely into the ventral portion of the thecal sac. A carbon dioxide laser was then used to shrink and subtotally resect the terminal portion of the lipoma emanating from the distal conus.

Pathological Examination. Histological analysis of the excised specimen revealed predominantly fatty and fibroconnective tissue, with small foci of skeletal muscle and atretic nerve twigs. Based upon these findings, the final pathological diagnosis of spinal cord lipoma (anatomical terminal-dorsal lipomyelomeningocele) was made.

Outcome. This patient remained neurologically normal with normal urological and anal sphincter function in initial postoperative follow-up and was enrolled in a comprehensive interdisciplinary spina bifida care program.

Discussion

Current understanding of lipomyelomeningocele formation implicates an error of primary neurulation that oc-
curs during the process of disjunction, when the neural tube separates from the surrounding ectoderm. It has been proposed that premature disjunction leaves the posterior neural plate open, permitting entry of migrating mesenchymal cells that are induced to form fatty tissue by the exposed neuroectodermal surface of the neural tube. Thus, spinal lipomas may consist exclusively of clusters of normal mature adipocytes separated by collagenous bands. However, careful histopathological analysis of lumbosacral lipomas may also reveal the presence of multiple different tissues derived from any of the 3 germinal layers, including smooth and striated muscle, neural and glial tissue, cartilage, lymphatic nodules, renal glomerules, endometrial tissue, and respiratory or intestinal epithelia.

It has therefore been suggested that lumbosacral lipomas may represent a form of benign teratoma. Despite extensive speculation, the origin of sacrococcygeal teratomas remains unclear. The traditional theory of teratoma formation is that totipotential primordial germ cells from the yolk sac migrate aberrantly to form germ cell tumors, including teratomas, instead of migrating to the gonadal ridge where they normally differentiate into the gonads, or alternatively that diffusely migrating germ cells are sustained in extragonadal locations due to abnormality in the regional distribution of growth factors or cell surface markers. This theory is supported by the observation of XX Barr bodies in the nuclei of some teratomas removed from male patients, and of immature teratomas that include derivatives of the embryonic yolk sac or of extraembryonic trophoblast tissue.

Alternatively, congenital teratomas may arise as the result of an inherited regional tendency to developmental error affecting the caudal embryonic segments. Several pieces of evidence support this alternative possibility. First, the embryonic caudal cell mass, consisting of residual undifferentiated mesenchymal cells from the primitive streak and Hensen node, is able to form all 3 germ layers both in vitro and in vivo. Second, there are several reports suggesting an association between spinal teratomas and dysraphism, including diastematomyelia. Third, although extragonadal teratomas are found most commonly in the sacrococcygeal region, the traditional germ cell theory does not necessarily account for teratomas found in certain other midline regions such as the mediastinum or diencephalon. Finally, as noted above, the observation that multiple different tissue types can be found within intraspinal lipomas, as in the case reported here, suggests that pluripotent cells may also give rise to teratomas in response to the regional trophic effects of developmental errors affecting the caudal embryonic segments.

The incidence of intradural spinal lipoma is estimated at 1 in 4000 births in the US, with a nearly 2:1 female predominance, although this is only approximate since the lesion is occult and may go undetected if asymptomatic. Sacrococcygeal teratomas are comparatively rare, despite being the most common congenital tumor in neonates, with an estimated incidence of 1–2 per 40,000 births and a 4:1 female predominance. Familial forms of both lipomyelomeningocele and sacrococcygeal teratoma have been reported, albeit infrequently. Neuronal tube defects occur at a similar rate in children born to families with a history of lipomyelomeningocele (4%) or of open neural tube defect (2–5%). Together, these data suggest that a genetic predisposition may play a role in certain cases of lipomyelomeningocele and of sacrococcygeal teratoma.

To our knowledge, this is the first reported occurrence of lipomyelomeningocele and sacrococcygeal teratoma in siblings. Considering that both conditions are relatively uncommon and that familial forms of each condition have been reported previously, it is less likely that these 2 related cases occurred coincidentally. Instead, we propose that these cases may be attributable to a single underlying heritable predisposition, implying that lipomyelomeningocele and intraspinal teratoma may in some cases represent different manifestations of a common underlying mechanism involving a regional developmental error affecting the caudal embryonic segments. Our observation does not speak to whether spinal teratomas always or even commonly form as a result of regional developmental error as opposed to aberrant germ cell migration or other alternative mechanisms.

Disclosure

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper. No portion of the contents of this manuscript has previously been published.

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