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# Intraspinal Tumor with Lumbosacral Agenesis

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Lumbosacral agenesis is an extremely rare congenital anomaly of the spine. We report such a case confirmed by metrizamide myelography and computed tomography (CT) after plain radiography of the spine. As a result of this extended investigation, an intraspinal lipoma was discovered, which was then excised completely.

## Case Report

A mature male infant was born to a 35-year-old gravida 6, para 4 mother who had gestational diabetes and was treated with 20 U of insulin daily. Although the infant tolerated normal feedings without regurgitation, he was continuously passing watery stools without any apparent sphincter control. On physical examination he was an alert child with stable vital signs. However, an erythematous firm lump measuring 3 × 3 cm with a longitudinal dimple was observed in the mid-upper lumbar area, without any exudate or bleeding. The arms appeared normal, but there was flaccid paralysis of the legs, which were hypoplastic with arthrogryposis; contractures of the hips and knees; and bilateral talipes equinovarus. Severe motor deficit was noted below the L1 level. Moreover, the child had a neurogenic bladder with continuous dribbling of urine.

Radiography of the lumbosacral spine (fig. 1A) showed complete absence of the sacrum and lumbar vertebrae below the L1 level. The pelvis was dysplastic with direct apposition of the iliac wings. A lumbar myelogram with metrizamide (fig. 1B) revealed a nontethered conus medullaris at the T11 level, while the truncated cauda equina ended at the T12-L1 level. At this lower level the metrizamide column was deformed and terminated abruptly. An extradural mass could not be excluded at this level.

To resolve the possibility of a mass, a limited CT scan within 4 hr of the myelogram showed a small, well defined mass at the L1 level, extending laterally on the left side and also posteriad to the dural sac, with slight flattening of the lowermost part of the sac. The attenuation value of the mass varied from -11 to -21 (fig. 1C), suggesting the presence of an extradural lipoma, epidural fat, or a dermoid tumor.

Surgical excision after T12 and L1 laminectomies revealed a large extradural lipoma that was completely removed. Histologic examination showed attachment of this neoplasm to the dura but not to the conus or nerve roots, with tissue fragments containing areas of fat necrosis and granulomatous cells. The midline dimple disclosed an inflamed dermal sinus contiguous with the terminal dural sac at the upper L1 level.

## Discussion

Lumbosacral agenesis involves the developmental absence of the sacrum and a varying number of lumbar vertebrae. Although the first case of sacral agenesis was reported in 1852 [1], not until more than a half-century later was lumbosacral agenesis first recognized [2]. Moreover, while sacral agenesis is rare, with about 400 cases described in a review of world literature [3], fewer than 50 cases of lumbosacral agenesis have been cited [e.g., 3-6].

Because ossification of caudal vertebrae occurs before the 10th week of fetal life, the pathogenesis of lumbosacral agenesis is assumed to operate during early gestation [7]. Both external and internal mechanisms have been postulated. The earliest theory in 1910 by Friedel [2] stated that minute embryonic trauma kinked the longitudinal axis to suppress caudal development. In another theory Freedman [8] suggested biochemical stresses may interrupt tissue differentiation in early embryonic life. Passarge and Lenz [9] and Sarnat et al. [10] have associated diabetes from the mother or other close relatives with an increased incidence of lumbosacral agenesis. Maternal diabetes was present in our case. Environmental factors have been proposed, ranging from xylene and gasoline [6] and insulin injection [11] to pyrexia [12] and serious illness such as rubella [13].

Since lumbosacral agenesis involves the absence of multiple sacral segments, these patients have neurogenic bladder and sphincter dysfunction [14]. Although such clinical findings may be isolated, other orthopedic and neurologic abnormalities are frequent. Dysplastic vertebrae and ribs, clubfeet, spina bifida, congenital hip dislocation, scoliosis, leg atrophy from paralysis, as well as visceral abnormalities such as imperforate anus have been reported [15]. Neurologic deficits include urinary incontinence and less often fecal incontinence, atrophy of the buttocks and leg muscles, and sensory changes. The latter findings are less frequent than motor dysfunctions and do not correlate well with the level of vertebral deformities [15].

Recently intraspinal neoplasms have been noted in sacral agenesis, including two lipomas and a dermoid cyst of the

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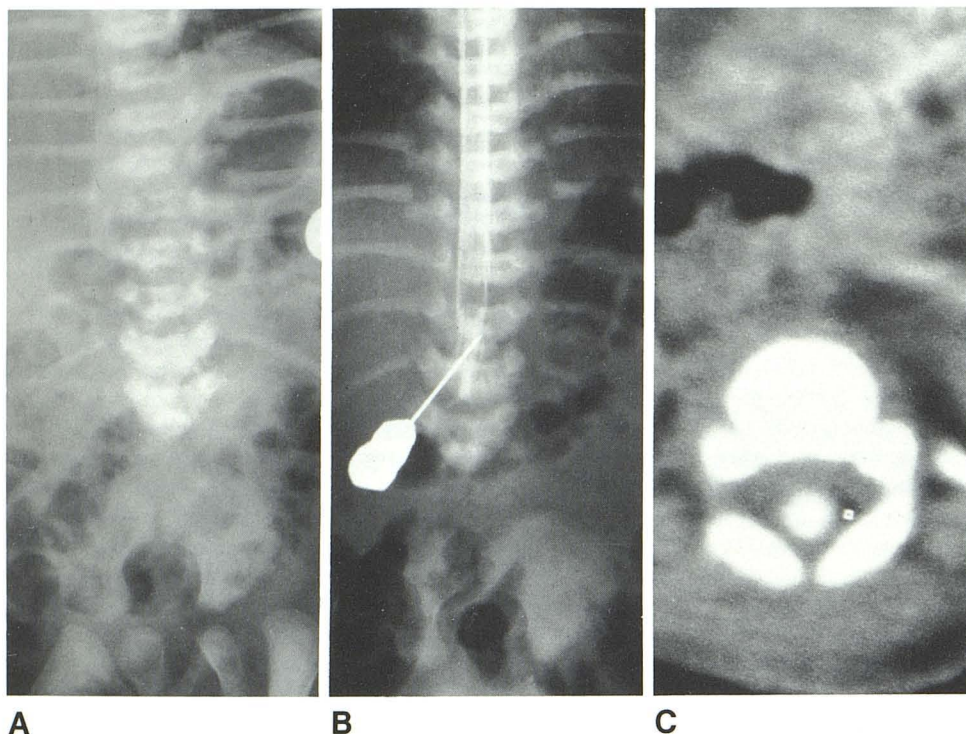


Fig. 1.—A, Conventional anteroposterior view of lower spine and pelvis. B, Metrizamide thoracolumbar myelogram in anteroposterior projection shows conus at T11 and cauda equina terminating at lower margin of T12. C, CT scan of hypodense mass adjacent to dural sac at superior level of L1. Low-density area has

cauda equina [7, 16], a lipoma attached to the dura and two lipomas within the conus medullaris [17], and another case report of an epidural lipoma beneath the conus [18]. Myelography has rarely been attempted with sacral agenesis, although it was successful in demonstrating an extradural mass (lipoma) not suspected on plain films [18].

Metrizamide myelography can be performed safely in most cases of lumbosacral agenesis to rule out an associated tumor. Myelography does not always define such tumors, as in the case report of an infant with caudal agenesis, in whom a large dermoid cyst embedded in sacral nerve roots was not identified by myelography, but instead was unexpectedly discovered during surgery [7]. Moreover, lumbosacral deficiencies of the subarachnoid space and the lower spinal elements can cause misleading interpretations of the myelograms. In such instances, CT may be useful in distinguishing defective dural sacs from an actual tumor. Without early radiographic confirmation, a delay in the diagnosis and treatment of intraspinal neoplasms may lead to increased disability of these patients [19].

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