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Sacral Agenesis Associated with Spinal Cord Syrxinx

Key Words

Sacral agenesis
Caudal regression syndrome
Hydromyelia
Syringomyelia
Magnetic resonance imaging

Abstract

The antemortem diagnosis of syringomyelia in the setting of sacral agenesis has not been reported before. We describe the clinical and neuroimaging features in 3 patients. None has required surgical intervention to date.

Sacral agenesis is associated with congenital anomalies of the spine and spinal cord, and the classical neuroimaging features have been described [1-3]. The antemortem recognition of syringomyelia in the setting of sacral agenesis has not been reported before. We present 3 cases. These cases were identified from a review of an institutional series of 22 patients with sacral agenesis/dysgenesis, 10 of whom have undergone magnetic resonance imaging (MRI) to date. The neuroimaging of sacral agenesis and sacral dysgenesis is the subject of an ongoing review at our institution, but because of the unanticipated incidence of syringomyelia of the thoracic cord, we present these cases as a separate report.

Case Reports

Case No. 1

A 3.15-kg (6-lb 15-oz) male was born at term to a mother with insulin-dependent diabetes mellitus. Bilateral talipes equinovarus was noted. Braces were placed at 6 months, and the child commenced walking at 18 months with obvious delay in gross motor milestones. Plain radiographs of the spine showed the absence of S2 through the coccyx. Examination proved distal extremity symmetrical atrophy and mild plantar flexion weakness, absence of ankle reflexes but preservation of normal sensation. Neurogenic bowel and bladder were obvious with no bowel control and a spastic dribbling bladder. The patient returned at 5 years of age with increased toe walking and complaints of bilateral lower extremity pain. Objective examination remained unchanged. MRI (fig. 1) of the thoracic and

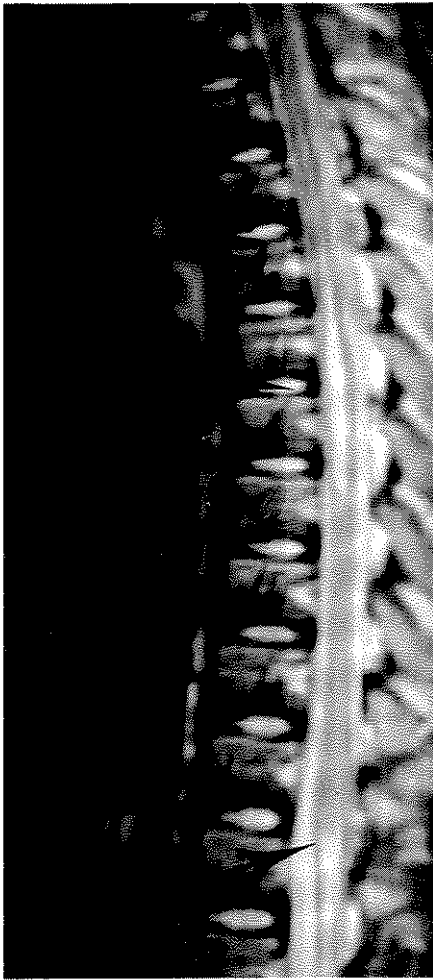
lumbosacral spine revealed a bluntly ending conus medullaris at T12 and a small thoracic syrinx. The craniocervical region was not imaged. The lower extremity pain resolved without specific treatment, and the patient has been followed for 3 years with no change in examination.

Case No. 2

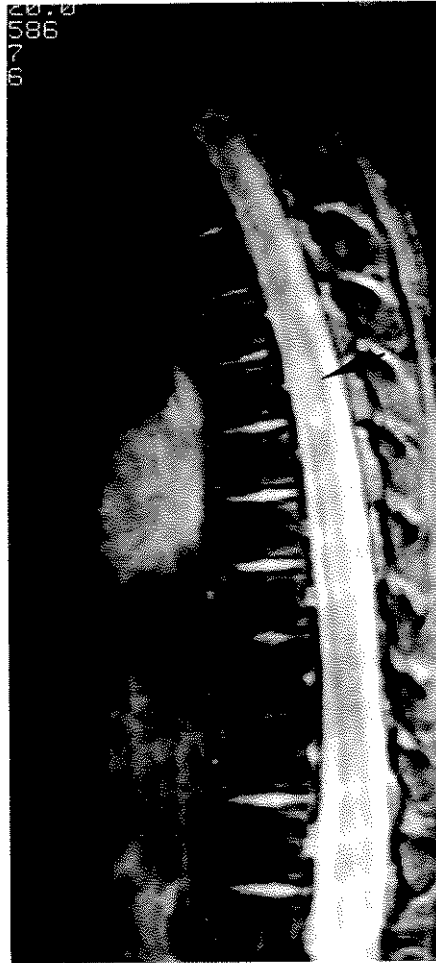
A 2.6-kg (5-lb 12-oz) male was delivered by cesarean section at 33 weeks of gestation because of fetal distress. The mother had gestational diabetes mellitus. The child walked at 18 months. He never achieved bowel or bladder control. Examination at 6 years of age revealed a palpable step-off at the sacrum with no cutaneous stigmata and a normal neurological sensorimotor examination. Anal sphincter tone was normal, although contractility was diminished. He had symmetrical lower extremity reflexes. Plain radiographs revealed complete absence of S3 through the coccyx. MRI (fig. 2) revealed blunt termination of the conus at L1 and a thoracic syrinx from T5 to T7. The remainder of the vertebral and spinal cord imaging was normal. There was no evidence of a Chiari malformation. The patient has remained neurologically unchanged over the past 2 years.

Case No. 3

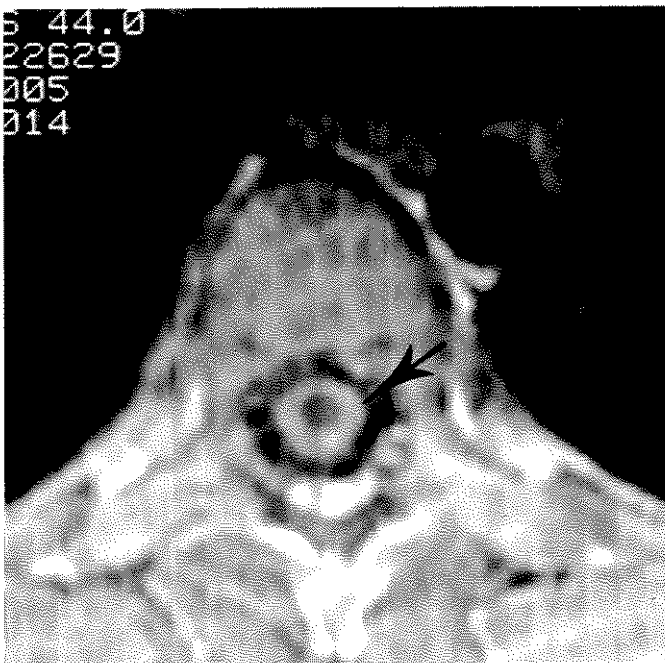
A 2.7-kg (6-lb) male was born by cesarean section. There was no history of maternal or familial diabetes. The patient was referred for neurosurgical evaluation at 9 months of age. Examination revealed imperforate anus and calcaneovalgus deformities of both feet. The patient had multiple cardiac anomalies including a unicameral ventricle and pulmonary artery stenosis. Neurological examination revealed no ankle plantar flexion power, but toe flexion and ankle and toe dorsiflexion were preserved. The patient had sacral dysgenesis with right hemisacral malformation, and MRI showed a blunt wedge-shaped conus at the L2 level with a thoracic syrinx at the T7 level (fig. 3). There was no Chiari malformation. There has been no clinical change in 1 year of follow-up.



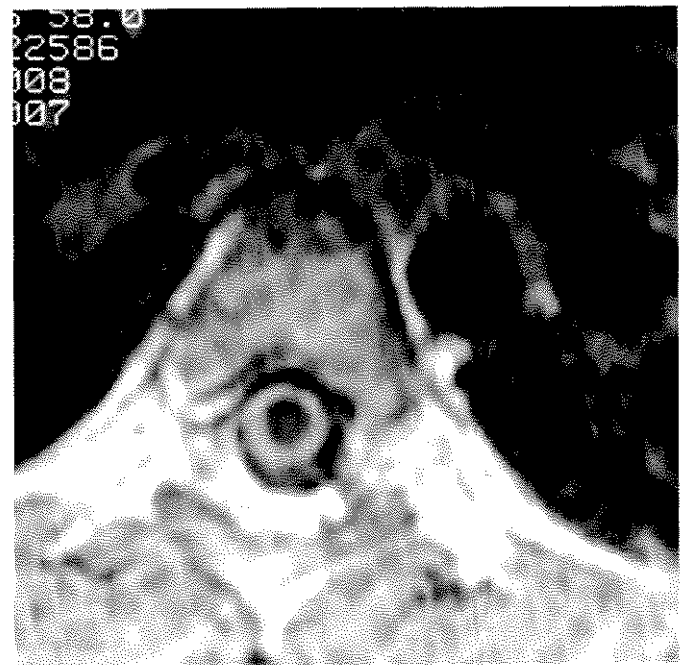
1a



2a



1b



2b



Fig. 3. Sagittal T1 weighted image with intramedullary cyst at the T7 level. No evidence of Chiari malformation.

Fig. 1. **a** T2 weighted sagittal image demonstrating intramedullary lesion of cerebrospinal fluid intensity and blunt conus. **b** Transaxial T1 image at eighth thoracic level demonstrating same intramedullary lesion.

Fig. 2. **a** Sagittal T2 image with CSF intensity intramedullary lesion centered at T6. **b** Unenhanced transaxial T1 image showing same lesion as in figure 2a.

Discussion

Sacral agenesis is associated with a potential for urological, neurological and orthopedic dysfunction. Over 200 cases have been reported in the literature. It is suggested that sacral agenesis occurs in 1% of infants of diabetic mothers [1, 4]. The radiological findings can vary from simple segmental distal sacral deformity or absence through lumbosacral vertebral element absence. Until relatively recently the neurological dysfunction, including urodynamics and sexual dysfunction, was thought to be static; however, cases of neurological decline and subsequent stabilization or amelioration following neurosurgical intervention have now been reported [2, 3]. Various forms of spinal cord tethering associated with sacral agenesis were described in these reports including tight filum terminale and intradural lipoma. These concomitant pathologies were responsible for neurological decline and were the target of surgical intervention. Most lesions that cause spinal cord tethering can be detected by MRI, and thus MRI may be of value as a screening tool to identify patients with sacral agenesis who are most likely to deteriorate neurologically.

In a postmortem study of a patient with sacral agenesis, Williams and Nixon [5] defined a diastematomyelia extending caudally from T8 for 5 cm. There was hydromyelia in the right hemicord and rostral to the cleft [5]. Neither diastematomyelia nor hydromyelia have been reported since. Postmortem studies of the spinal cord in sacral agenesis have documented many other anomalies, such as meningocele, adhesive arachnoid bands, myelomeningocele, absence of ventral and dorsal spinal cord fissures, fusion of ventral horn grey matter and preterminal central canal dilatation or forking of the central canal [6-9].

The pathophysiology of the syringomyelia associated with sacral agenesis is obscure. It is possible that syringomyelia is a developmental phenomenon related to abnormal patency of the central canal, in which case it would be more correctly termed 'hydromyelia'. It is also possible that the syringomyelia represents an injury of the cord on the basis of tethering. Further investigation of this possibility awaits objective techniques for noninvasive assessment of mechanical tension in the spinal cord [10]. Developmental and mechanical factors may work together.

The clinical significance of our findings is difficult to predict. Only a single patient has had long-term follow-up, and he has not deteriorated neurologically; however, the potential for decline from an enlarging syringomyelic cavity is well documented in other diseases. In the long-

term management of patients with sacral agenesis, issues of skeletal stability, bladder function, and neurological function must be assessed periodically in a systematic fashion, and at our institution such patients are followed through a multidisciplinary clinic for congenital spinal anomalies. Baseline spine radiographs, urodynamic investigations, and MRI are obtained. Somatosensory evoked potentials have been reported to be useful in the management of children with congenital spinal cord disorders as well, but this modality has not been used rou-

tinely in our clinic [11, 12]. Spine radiographs are repeated annually for patients with scoliosis. Urodynamic evaluations are repeated only when indicated for evaluation of new symptoms. MRI is repeated annually for patients with syringomyelia. Progressive scoliosis, interval enlargement of the syringomyelic cavity on follow-up imaging, and new pertinent disturbances of neurological function or bladder function are all indications for syrinx drainage, but none of these problems has been encountered to date in our patients.

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