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Agensis and Dysgenesis of the Sacrum: Neurosurgical Implications

Key Words

Caudal regression
Lipomyelomeningocele
Magnetic resonance imaging
Occult spinal dysraphism
Sacral agenesis

Abstract

We reviewed 27 patients with congenital anomalies of the sacral spine. There were 16 males and 11 females with a mean follow-up of 81.1 months (range 8-211 months). Fifteen patients had sacral agenesis and 12 had sacral dysgenesis. Fifteen patients had neuroimaging of the spine. Seven patients had conus termination below the L2 vertebral body. Four patients had associated thoracic syringomyelia and 6 patients were identified with caudal or dorsal lipoma. There were only two episodes of neurological deterioration, both in a single patient who had a lipomyelomeningocele, in 182 patient-years of follow-up. Four patients with low lying conus had preemptive spinal cord exploration for release of tethering in order to prevent neurological deterioration. Patients with agenesis or dysgenesis of the sacrum should undergo magnetic resonance (MR) imaging of the spine in order to detect spinal cord lesions associated with progressive neurological deterioration. Findings on MR imaging are more likely to correlate with clinical course than findings on skeletal radiography.

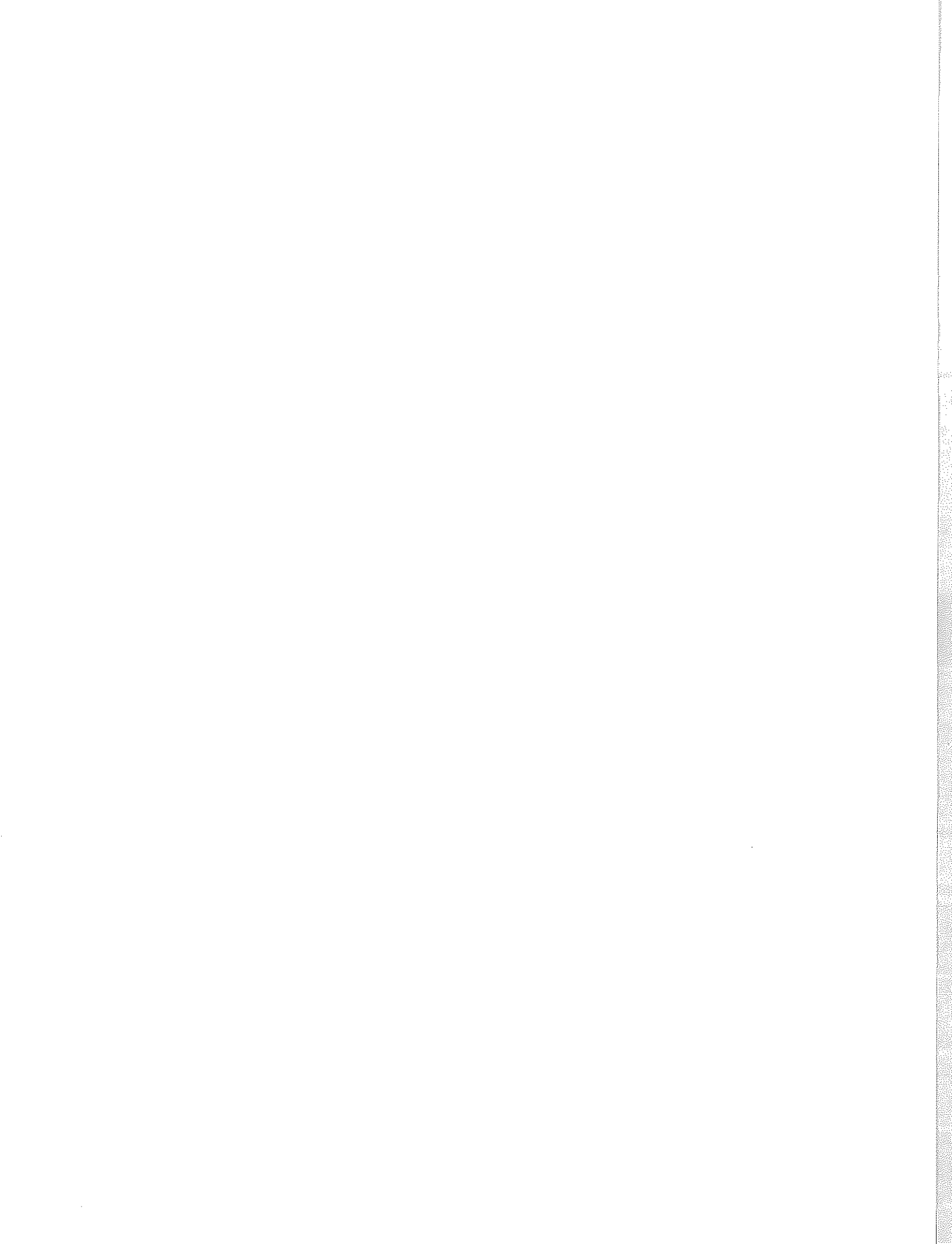
Introduction

The role of the neurosurgeon in the management of patients with sacral agenesis and sacral dysgenesis has been problematic. Many authors have reported the neurological deficits associated with these conditions to be static [1-3]. However, recent publications have described progressive neurological syndromes arrested or reversed by neurosurgical treatment [4-11]. To some degree, these inconsistencies may be accounted for by the inadequacies of current nosological schemes. Pang and Hoffman [4] initiated the concept that there was potential for progressive neurological decline in their early treatise on sacral agenesis. A recent report has stressed a higher incidence of clinical deterioration than previously thought in patients with sacral agenesis, particularly in patients with tethered cord

or dural stenosis [5]. We have reviewed our institutional experience with sacral agenesis and sacral dysgenesis to obtain a better sense of the incidence of neurological deterioration in these conditions and to identify whether modern neuroimaging techniques might support a more refined system for classification of congenital anomalies of the sacrum.

Methods

Indexed diagnoses of congenital lumbosacral anomalies from 1970 to 1993 at the Oregon Health Sciences University Hospital and at the Shriners' Hospital for Crippled Children Portland Unit were sought by computerized record review. *Sacral agenesis* was defined as symmetrical congenital absence of one or more of the most caudal lumbosacral vertebrae, and *sacral dysgenesis* was defined as asym-



metrical deformity in this same region. Patients with isolated sacral spina bifida or coccyx agenesis were not included. Anomalies involving elements of the lumbosacral spine but sparing the most caudal segments were excluded. For example, one patient with hemivertebra at S1, a low lying conus, a terminal lipomyelocystocele and a thoracic syrinx was excluded because the sacral vertebral segments below S1 were normal except for spina bifida.

The term *caudal regression syndrome* is used to describe a constellation of congenital anomalies related to a disturbance of the caudal cell mass and the precursors of the urogenital system and the lower intestinal tract, to which it relates closely in the late embryonic period. In addition to sacral agenesis and dysgenesis, common features are anal atresia, cleft lip and palate, renal dysplasia, and lower extremity abnormalities ranging from short femora and absent fibulae to sirenomelia [12].

Records were reviewed for gestational and family histories, physical findings on general and neurological examination, and clinical course. Operative reports were studied for descriptions of gross pathology. Inclusion in this study was based on skeletal radiography, but correlates with myelography and magnetic resonance (MR) imaging of the spine were sought. Images were specifically reviewed for the level and shape of the conus medullaris, the presence of syringomyelia, lipoma, spina bifida, meningocele or associated pathology.

Case Reports

Case 1

A white male infant, 3.8 kg, was delivered by uncomplicated cesarean section at 38 weeks gestation. The mother had gestational glycosuria with poor weight gain in the first two trimesters. Prenatal fetal ultrasound showed bladder obstruction and omphalocele. Amniocentesis revealed no chromosomal abnormality. Multiple urogenital anomalies noted at birth included omphalocele, cloacal exstrophy, ectopic imperforate anus, renal (fused) ectopia, undescended testes and left lower extremity atrophy and malformation. Multiple corrective procedures were performed for these anomalies. The patient was first referred for neurosurgical review at 8 months of age with a question of less movement in the left lower extremity. Examination revealed a strawberry nevus over the sacral region extending to the left leg with external rotation of that extremity and flexion contractures with atrophy below the knee. The patient had left-sided L4 motor and sensory levels with an absent left achilles reflex. The right leg neurological examination was normal, and anal tone was diminished. Plain spine radiographs revealed sacral dysgenesis and an MR imaging demonstrated a lumbosacral lipomeningocele and a conus syrinx with a low lying conus (fig. 1). The patient had operative exploration with subtotal excision of the lipoma, syrinx shunting, and release of the tethering. There was some demonstrable clinical improvement postoperatively with increased spontaneous movement in his left leg and return of sensation to S1.

The patient presented to another neurosurgical center 2 years later having started walking with the aid of a walker and leg braces at 20 months of age, with static lower extremity function but worsening and persistent left lower extremity pain. Repeat MR imaging showed residual lipoma. Reexploration of the region with incomplete release of the cord tethering and resection of lipoma resulted in postoperative pain relief, and the patient has remained neurologically stable since.



Fig. 1. Sagittal T1-weighted MR imaging demonstrating conus termination at L3, conus syrinx, and large lipoma.

Case 2

A 3.2-kg male was born at term to a mother with insulin-dependent diabetes mellitus. Bilateral talipes equinovagum deformity was noted at that time. The patient was braced at 6 months and walked at 18 months. Distal extremity atrophy was present with mild weakness but complete preservation of sensation in both lower extremities. Neurogenic bowel and bladder dysfunctions were evident with no bowel control and a spastic dribbling bladder. The patient presented in 1988 at 5 years of age with complaints of lower extremity pain and increased toe walking, although his lower extremity strength and sensation appeared normal. There was no change in his bowel or bladder function. MR imaging revealed a thoracic syrinx and blunt termination of the conus with mild dorsal wedging at T12. No surgical intervention was recommended. His pain resolved without specific treatment, and he remained stable over 5 years of follow-up. Repeat MR imaging has shown no change in the syrinx. There was no evidence of Chiari malformation. This case has been reported before [6] (fig. 2).

Results

Twenty-seven patients were identified with congenital lumbosacral anomalies meeting the inclusion criteria (table 1). There was symmetrical absence of lumbosacral elements (agenesis) in 15 patients and partial, asymmetrical loss (dysgenesis) in 12 patients. There were 16 males and 11 females. Mean age at referral was 23 months (range birth to 11 years). Mean follow-up was 81.1 months (range 8–211 months). Ten patients exhibited severe mul-

