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Case Report

Caudal regression syndrome diagnosed after the childhood period: a case report

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ABSTRACT

Caudal regression syndrome is a congenital syndrome that presents with pathology of spinal tract migration during the embryologic period. In this paper, we report a 16–year-old caudal regression syndrome case. This syndrome is very rare, especially after the childhood period. In this patient, caudal regression syndrome was demonstrated by imaging techniques. The patient was planned to be followed without operation. Treatment methods and the follow- up period in caudal regression syndrome are discussed. ***Neuroanatomy; 2005; 4: 16–17.***

Key words [caudal regression] [spinal cord] [abnormality] [disease]

Introduction

Caudal regression syndrome is a pathology caused by anomaly of spinal trunk 'ending', and encompasses a wide range of anomalies of the hind end of the trunk, including partial agenesis of the thoracolumbosacral spine, imperforate anus, malformed genitalia, bilateral renal dysplasia or aplasia, pulmonary hypoplasia, and in the most severe deformities, extreme external rotation and fusion of the lower extremities (sirenomelia). The syndrome is significantly associated with several systems, including congenital cardiac disease (24%), genito-urinary disease like hydronephrosis, renal agenesis, epispadias and hypospadias (24%), orthopedic anomalies like gluteal anomalies, scoliosis, and talipes deformities (12%), and progressive deficits like back and leg pain (30%). Tethered cord, dermoid cyst, lipoma and diastematomyelia may emerge in the central nervous system [1-3].

Examination and clinical follow-up are important for the decision of operation if these anomalies are suspected to cause neurological deficits. Myelography and myelo-computerized tomography (CT) have been replaced today by magnetic resonance imaging (MRI) as the gold standard for diagnosis [1, 2, 4-6]. There is a definite but incomplete association of the syndrome with diabetes mellitus; 1% of the offspring of diabetic mothers will have a form of this syndrome. Genetic changes caused by teratogens and pathologies 7q have been suggested as other factors [2].

Caudal regression syndrome has been reported only rarely after the childhood period in the literature.

Case Report

A 16-year-old female was examined for back pain while on follow-up for neurogenic bladder by the Urology Clinic. There was no symptom other than urinary incontinence, and no history of another illness. Family history revealed that the mother of the patient had diabetes mellitus before and during her pregnancy. Lumbosacral MRI scan demonstrated termination of the spinal cord at the T11-T12 level and narrowing of the canal at the L4-L5 level, and the patient was diagnosed as caudal regression syndrome.

No fracture lines were found at bone structures, but the partial sacral agenesis was shown on direct films (Figure 1). The spinal cord fit the classic caudal regression syndrome imaging by lumbar MRI (Figure 2).

Detrusor hyperreflection was shown with cystometrogram (CMG) and minimal residue was observed on voiding cystourethrogram (VCUG). The patient's complaints significantly improved with anticholinergic medication. No operation was planned because the patient improved with medications and because no other pathological signs were determined. There has been no progression of the patient's complaints after three years of follow-up.

Discussion

Caudal regression syndrome is generally diagnosed in the early years of life and requires surgical intervention

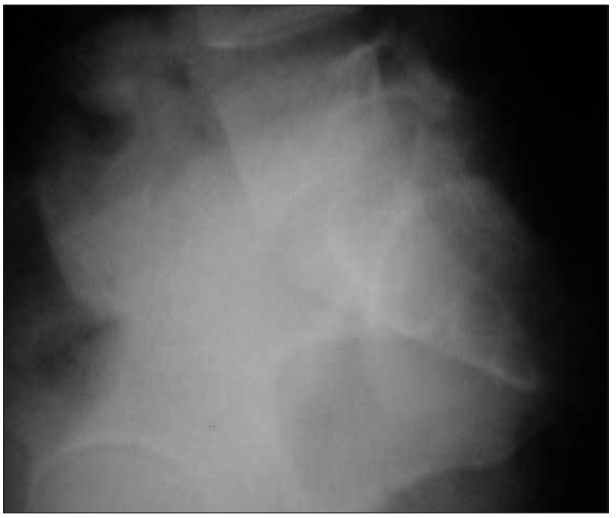

Figure 1. Lateral radiograph revealing a partial sacral agenesis.

in the case of neurological deficits. The signs on neurological examination may range from a variety of minimal deficits to severe paralysis. Motor deficits are generally more severe than sensory deficits.

The syndrome has been shown to occur more frequently in the offspring of diabetic versus non-diabetic mothers. Although hyperglycemia in the early stages has been implicated, the pathogenesis remains unknown. Trauma, nutritional problems, toxic agents and genetics are the other factors suggested in the etiology [3, 4].

Myelography and myelo-CT were previously used for diagnosis. Sacral agenesis and vertebrae anomalies that are shown by direct films may give information about the syndrome. Distal vertebral anomalies and fetal spine anatomy may be seen by obstetric ultrasonography (USG), and in the intrauterine period, amniofusion may be important, especially in cases associated with oligohydramnios. The superiority of lumbosacral MRI is generally accepted today [2, 5, 6].

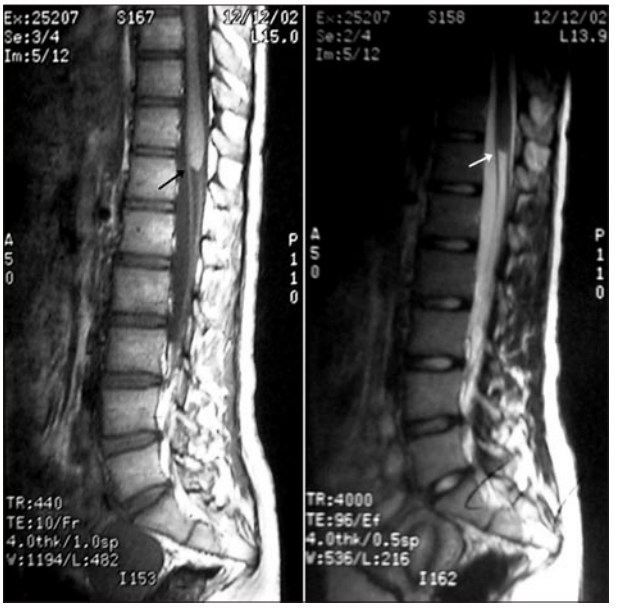

Figure 2. Caudal regression syndrome. Sagittal T1-weighted (left) and T2-weighted (right) midsagittal MRIs. Partial sacral agenesis, with only S1, S2 and a portion of S3 present. The conus lies at T12 and shows bulbous angulated termination (arrows). The distal bony canal and thecal sac are narrow.

Patients may require surgical intervention for decompression and vertebral anomalies, especially in caudal regression syndrome with neurological deficits. Some authors have advised that in tethered cord cases in caudal regression syndrome, cutting of the filum terminale would be useful to prevent secondary infection caused by residual urine in the bladder [7].

We suggest clinical follow-up after the childhood period for those patients with no severe or progressive neurological deficit, as in our case. However, patients with severe or progressive neurological deficits should be operated.

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