

Case Report

CAUDAL REGRESSION SYNDROME

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ABSTRACT

Caudal regression syndrome (CRS) is a rare neural tube defect affecting terminal spinal segments and cord manifesting as neurological deficit ranging from bladder and bowel involvement to severe sensory motor deficits in lower limbs. It has sporadic appearance and maternal diabetes, genetic factors, teratogens and hypoperfusion are considered as possible etiologic factors and it can be associated with other congenital anomalies. It is generally diagnosed by antenatal sonography or in early childhood. In this paper we report a case of caudal regression syndrome diagnosed at age of 12 yrs with minimal neurological deficits.

Key Words: *Lumbosacral Agenesis, Sirenomelia, Urinary Incontinence*

INTRODUCTION

Caudal Regression Syndrome or syndrome of caudal regression is characterized by abnormal development of the caudal aspect of bony spine and cord of the developing fetus resulting in neurological deficits. A wide range of abnormalities may occur including partial absence of sacrum causing no apparent symptoms to extensive deformity of the lower vertebrae, pelvis and fused lower limbs (sirenomelia). It is predominantly associated with other genitourinary abnormalities with cardiac and tracheo-oesophageal malformations in severe cases. The intelligence is normal and curative treatment of the condition is absent and supportive care is all that is available.

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A 12 year old girl visited our out patient Department of Pediatrics with primary complains of absence of urinary control since birth and dribbling of urine with foul smell. Minimally altered gait was noted and no history of any other systemic illness could be obtained. Pathological examination of urine revealed plenty of pus cells and 1-3 epithelial cells per HPF. Urine culture revealed E.Coli with sensitivity of third generation cephalosporins.

A skiagram of lumbosacral spine was done and it revealed malformed sacrum (Fig. 1). CT scan pelvis with 3D (SSD) reconstruction (Fig. 2) reveals significantly small ala of sacrum bilaterally and only 2 segments of vertebral bodies identifiable. The distal sacrum and coccyx were absent. The iliac bones including acetabulum and head of femur appeared normal. A Magnetic Resonance Imaging (MRI) scan was performed and it revealed sacralized L5 vertebra fused with malformed sacrum and absence of coccyx. Conus was blunt and wedge shaped terminating at D11-12 level (Fig 3) and no tethering or syringomyelia seen. No dural defect or evidence of aberrant fat tissue in spinal canal was noted. The urinary bladder was marginally aberrant in shape with irregular thickening of wall and small diverticulae. The anal canal and distal rectum did not reveal any significant abnormality (Fig 4). No hydronephrosis or hydroureter was noted. Echocardiography was done and it did not reveal any developmental cardiac abnormality. The patient was treated with antibiotic (ceftriaxone 100 mg per Kg per day) to clear urinary tract infection. After treatment with antibiotics no improvement in urinary incontinence was observed and hence prescription of anticholinergic medication was considered (oxybutynin 0.2 mg per Kg per day). On follow up after 3 months, there was no progression of patients complains.

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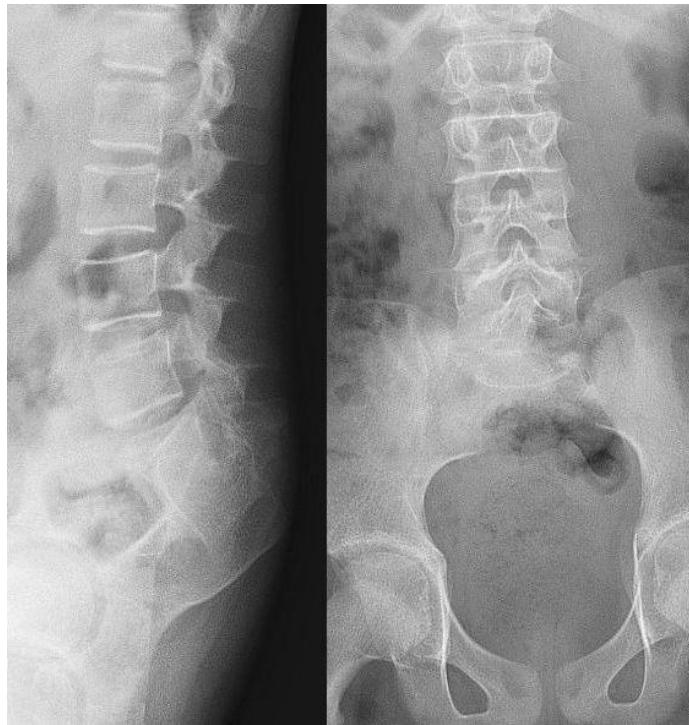


Figure 1: X-ray lumbosacral spine AP and Lat view showing abnormally short sacrum.



Figure 2: 3D surface shaded display (SSD) of bones obtained from axial CT scan images showing small ala of sacrum predominantly on left side with only two vertebral bodies.

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Figure 3: T2 weighted MRI sequence of whole spine in sagittal plane revealing wedge shaped terminal end of conus with no syringomyelia or tethering. L5 vertebra is completely sacralized and aberrant sacrum is noted.

DISCUSSION

Caudal regression syndrome comprises of a spectrum of developmental anomalies including caudal most vertebrae and cord, neurological deficits, sirenomelia, anal atresia, malformed external genitalia, exstrophy of bladder, renal abnormalities including ectopia and aplasia, rectovesical and rectoureteral fistula, pulmonary hypoplasia and cardiac abnormalities [Barkovich 2005] [Naidich 2009]. The least severe cases of caudal regression are characterized by partial deformation of the sacrum which is generally unilateral. The next level is indicated by a bilateral

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deformation. The most severe types involve a total absence of the sacrum and distal lumbar vertebrae. The exact etiology is unknown but appears to be due to intrauterine insult before 4



Figure 4: Sagittal T2 weighted image of pelvis showing abnormal thickening of wall of urinary bladder with few small diverticulae. Also noted are normal uterus, rectum and anal canal.

weeks of gestation which could be toxic, ischemic or infectious. Other possibilities could be injury to notochord or abnormality of glucose transporter genes. There is a definite association with maternal diabetes and a sixth of cases of caudal regression syndrome have diabetic mother. Proper control diabetes before conception and in first eight weeks of gestation significantly reduced the chances of the anomaly [Kassai 2008].

Detailed clinical examination and radiological investigations are essential for proper management of condition. Myelography and CT myelography has been replaced by MRI as gold standard [Kahilogullari 2005] as it can provide details of almost all the anatomical abnormalities associated with the condition. Lumbosacral hypogenesis is frequently associated with genitourinary and gastrointestinal anomalies named as OEIS (omphalocele, cloacal exostrophy, imperforate anus and spinal deformity) and VACTERL (vertebral anomalies, anorectal malformation, cardiac malformations, tracheoesophageal anomalies, renal agenesis and limb abnormalities) syndrome. Hypoplastic anomalous distal cord is seen in almost all case of sacral agenesis predominantly ventrally resulting in blunted wedge shaped terminal end of cord. Tethering of cord is a common associated anomaly more commonly seen in milder forms of hypogenesis and in such case the terminal cord is not blunted but elongated like typical tethered cord [Barkovich 2005]. Clinically, patients with milder abnormalities have motor and sensory deficits, sphincter disorder and almost always neurogenic bladder. In cases of sacral agenesis tip of conus located below L1 level is almost always suggestive of tethering [Pang 1993]. Progressive neurological complains

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is suggestive of tethering of cord which is seen in about 60% cases of lumbosacral agenesis. In severe cases aberrant fibrous tissue is noted at distal end of thecal sac with dural stenosis or narrowing of bony canal of caudal most intact vertebra, surgical repair of which leads to significant clinical improvement [Naidich 2009]. The treatment plane depends on degree of neurological deficits and hind limb abnormality. Muscarinic receptor antagonists are medical treatment of choice in cases of neurogenic bladder. It decreases intravesicular pressure and frequency, increases bladder capacity and alters bladder sensation. It reduces the tone of detrusor muscle by decreasing parasympathetic control [Brown 2011]. In severe cases with sirenomelia full amputation of legs with disarticulation of hip is done. Colostomy is done for imperforate anus.

To summarize, caudal regression syndrome is a rare congenital anomaly that represents a continuum of congenital malformations ranging from agenesis of the lumbosacral spine to the severe cases of sirenomelia with hind limb fusion and major visceral anomalies that can be detected by dedicated antenatal sonography and MRI is investigation of choice postnatally. No curative therapy is available and surviving patients managed according to the degree of clinical complains mostly due to neurogenic bladder and its complication of progressive renal damage and neuromuscular deficits.

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