

# Lumbosacral Agenesis

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**Key words :** - caudal regression syndrome, lumbosacral agenesis.

## Introduction :

Caudal regression syndrome is an uncommon malformation in the general population occurring in approximately 1 of 25000 live births. The term is commonly applied to a group of disorders characterized by absence of variable portion of the caudal portion of the spine. Hohl first described agenesis of the lower spine in 1852 and Friedel redescrbed it in 1910.



Fig 1



Fig 2

## A case report

A male child of 8 months was referred for radiological evaluation. He was first sibling of 24 yr old diabetic mother. On examination there was a hump in the lumbar region with bilateral gluteal dimples. Normal convexity of

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sacrococcygeal region was lost . Anal opening was horizontal. Fixed calcaneal deformity of the feet was present. There was significant wasting of both lower limbs. Anteroposterior and Lateral Radiographs of LS spine revealed absence of L5 vertebra with upturned L4 vertebrae, sacrum was completely absent with apposition of both iliac blades with midline cleft between them. (fig 1,2 ). Diagnosis of lumbosacral agenesis was made . USG abdomen and pelvis done with 5 MHz linear probe showed left renal hypoplasia and right sided hydronephrosis. MRI was done with 0.5 Tesla gyroscan. T1 and T2 WTD images were taken in sagittal , coronal and axial planes, which showed complete agenesis of L5 vertebra and sacrum.(fig 3,4,5,6). Both iliac bones were seen articulating with each other below L4 with resulting shortening of transverse pelvic diameter.(fig5) Conus ended abruptly at the level of T11 and was chisel shaped. Distal thecal sac was seen tapering to a narrow channel ending at L4 level (fig3,4). There was no evidence of lipomyelomeningocele or hydromyelia or terminal myelocystocele. On coronal T2 WTD image, left renal hypoplasia and right renal hydronephrosis was confirmed.(fig6)

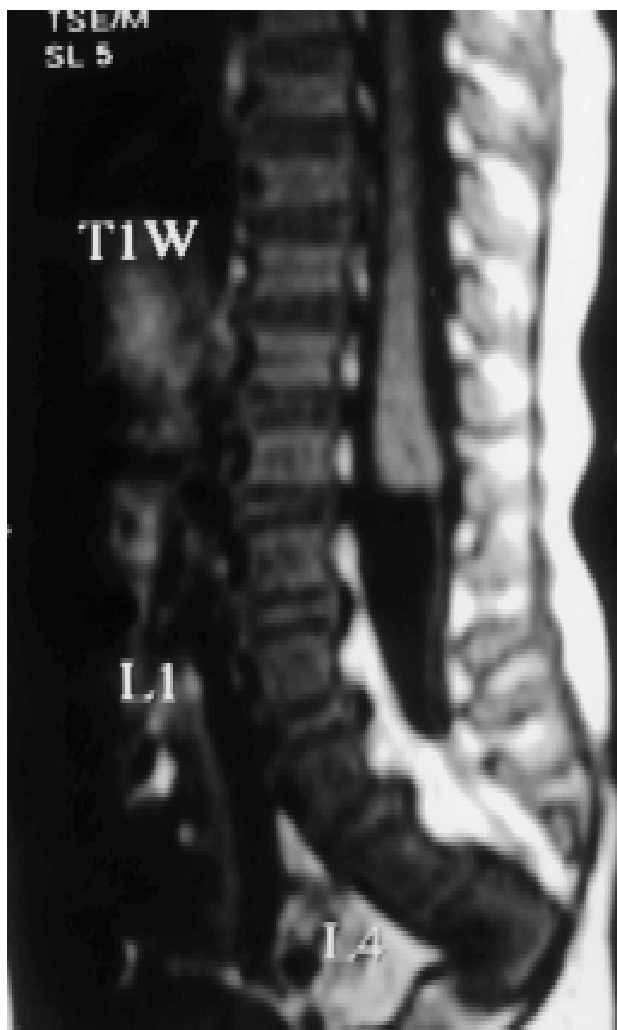
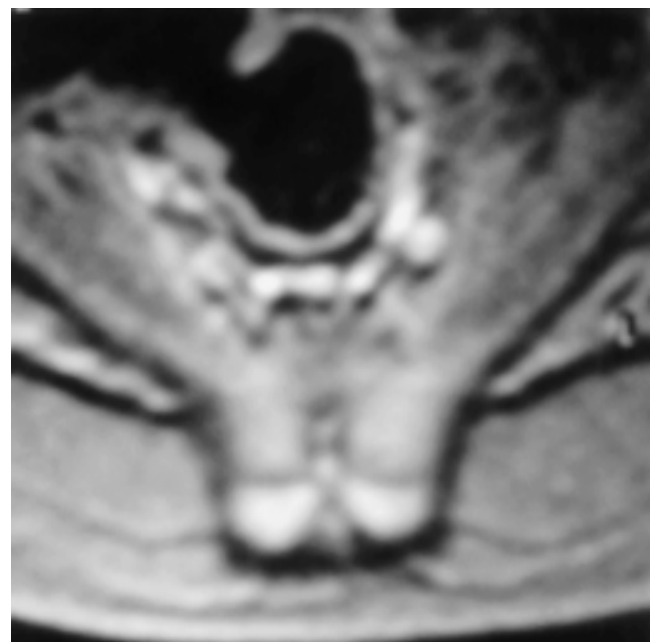
Fig 3 : T<sub>1</sub>W - SagittalFig 4 : T<sub>2</sub>W - Sagittal

Fig 5 : Axial

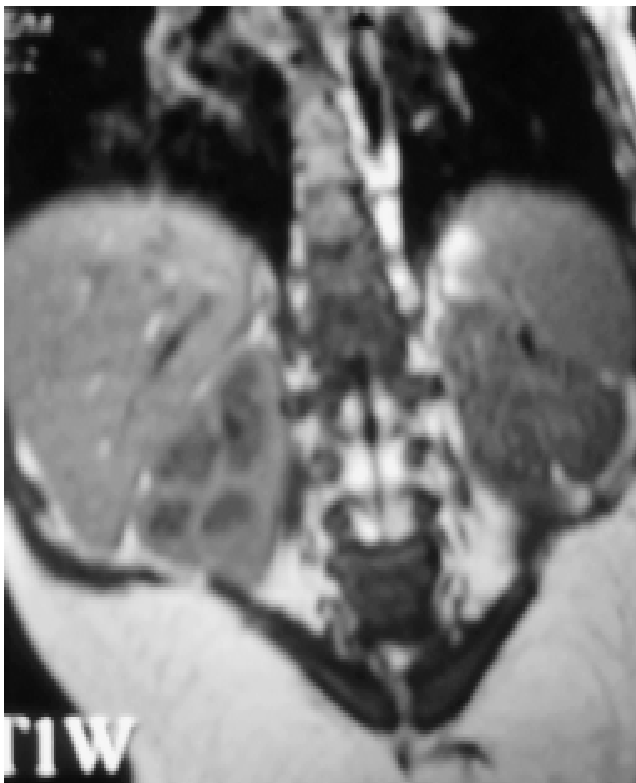


Fig 6 : T<sub>2</sub> - Coronal

## DISCUSSION

Lumbosacral agenesis is an uncommon condition characterized by absence of different segments of lumbar spine along with total or partial absence of sacrum.<sup>1,2</sup> The etiology of the syndrome is Unknown . it is possibly associated with maternal insulin-dependent diabetes (16% - 50%)<sup>1,2,6</sup>

Disturbances of the caudal mesoderm including the caudal cell mass and cloaca occur before the fourth week of gestation. Toxic, infectious or ischemic insults presumably impairs the normal migration of neurons, paraxial mesoderm cells (somites that form vertebra) and the lateral mesodermal cells that form the lower digestive tract.<sup>3</sup>

A dominant inheritant form of Lumbosacral agenesis has been shown to result from defects in the HLXB9 homeobox gene mapping to chromosome 7q36. This gene is also expressed in pancreas perhaps accounting for the association of lumbosacral agenesis with Insulin & Diabetes.<sup>6</sup>

Motor deficits are present and correspond to the level of vertebral agenesis. Function of quadriceps and the hip girdle is typically preserved unless there is concurrent lumbosacral dysraphism. Sensation is better preserved than motor function.<sup>3,6</sup> Thus total sacral agenesis may

be associated with complete motor paralysis below the quadriceps but relatively intact sensation in the perianal region. 6. Urinary and Bladder dysfunction are constant. 1,2,6 Associated problems as expected from embryology include OEIS complex (omphalocele, cloacal extrophy, imperforate anus, spinal deformities), VATER syndrome (vertebral anomalies, ano rectal malformation, tracheo-esophageal fistula, renal anomalies and limb anomalies), congenital heart defects (24%), genitourinary complaints with unilateral renal agenesis, hydronephrosis, pelvic and horseshoe kidneys, epispadias and hypospadias(24%); orthopedic deformities such as hip dislocation, flexion contractures, genu recurvatum, posterior compartment atrophy, telipes deformities and scoliosis (12%); progressive neurological deficits and back and leg pains(38%). females show uterine anomalies or rectovaginal fistulas. 1,2,3,6

On USG, the diagnosis of caudal regression syndrome has been established as early as 9 to 11 weeks gestational age, based on abnormal appearing yolk sac and short crown rump length. By 11 weeks , protuberance of caudal region was noted.<sup>4,5</sup> . The diagnosis of this syndrome is based on the presence of a variety of abnormalities of varying degrees of severity.<sup>4,5</sup> . Amniotic fluid is normal in most cases unlike in sirenomelia which is associated with profound oligohydroamnios.<sup>5</sup> Because the sacrum is not well ossified in the late first trimester or early second trimester not every case of caudal regression syndrome can be identified early, particularly if it is a mild form of the disorder.<sup>4,5</sup>.

As seen on MRI ,the position of the conus defines two distinct groups of patients with sacral agenesis. In group 1 (41%),the conus ends cephalic to the lower border of L1 vertebra. The conus is typically deformed (92%) and terminates abruptly at T11 or T12 as if the normal distal tip was absent. The distal central canal may be slightly dilated as a terminal hydromyelia. In this group the sacrum usually ends at or above S1.<sup>3,6</sup> Coronal T1 weighted images will show Christmas tree configuration formed by distal thecal sac, root sleeves and nerve roots within the epidural fat of the spinal canal.

In group 2 (59%) the conus ends lower, below L1, and is elongated, tethered by thick filum(65%),terminal myelocystocele(15%), transitional lipoma(10%) or elongated cord with terminal hydromyelia(10%). In these patients sacrum is relatively well preserved. Neurological deficit is more in patients with low tethered cords. 3,6

The orthopedic deformity depends on the extent of vertebral agenesis, the symmetry of involvement and whether the ilia articulate with the sides of the last intact vertebra(relatively wide pelvis) or with each other inferior to the last vertebra(narrow pelvis). Unilateral sacral agenesis leads to marked pelvic tilt and scoliosis.

Isolated agenesis of the coccyx is an incidental finding.6

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