Syringomyelia in Caudal Dysplasia Sequence

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Abstract
Caudal dysplasia sequence (CDS) comprises developmental anomalies of the caudal vertebrae, neural tube, urogenital and digestive organs, and hind limbs, the precursors of all of which are derived from the caudal eminence. Although the syndrome is well recognized, the etiology and pathogenetic mechanisms are poorly understood. We report syringomyelia in association with the CDS, which has not been described previously and recommends inclusion of this entity in the spectrum of abnormalities encountered in this syndrome.

INTRODUCTION
The clinical, radiologic and the radionuclide imaging features of a case of caudal dysplasia sequence (CDS) is described. The etiopathogenetic controversies and differential diagnoses of this rare and complex neural tube defect along with the continuum of congenital malformations are discussed. The corrective measures for the various malformation are highlighted. Association of syringomyelia with this congenital syndrome, not described previously, is documented in the present case.

CASE REPORT
A 2 kg full term male newborn was delivered vaginally to a 25 years nondiabetic primigravida. At birth, the child had an imperforate anus and a lumbosacral swelling (Fig. 1) through which a vertebral defect could be felt. The parents of the child were apparently healthy and did not have any major illness in the past. The gestation period was uneventful except for the fact that an obstetric ultrasonography done at 31 weeks of gestational age was suggestive of presence of segmentation anomaly involving the lumbosacral spine. It also suspected bilateral hydronephrosis and hydroureter. The imperforate anus was corrected by surgery. The child had motor and sensory neurological deficit with reduced power in all the muscles of the lower limb. At the age of two years he was able to sit only with support and also had impaired control over bowel movements and urination since birth. The child underwent intravenous urography (IVU) for renal evaluation which revealed nonvisualisation of right kidney with marked dilatation of collecting system of left kidney and markedly dilated tortuous left ureter (Fig. 2). Congenital segmentation anomaly of the lumbosacral spine was also noted. MRI of the lower dorsal and lumbar spine (Figs. 3 and 4) was performed in the sagittal and coronal planes using T1 and T2 weighted sequences. It revealed complete sacral agenesis with L3 representing last intact vertebral body. The cord was low lying and tethered to a lipomatous soft tissue which was contiguous with the subcutaneous fat of the back. Focal syringomyelia was noted at D12, L1. Right kidney was atrophic with compensatory hypertrophy of the left kidney and bilateral hydroureter. 99mTc-DTPA renogram (Fig. 5) showed prompt perfusion and good cortical uptake in the left kidney and absent perfusion and cortical uptake on the right side. From the sixth minute image onwards, another area of radioactivity appeared just below left kidney which gradually became intense and bigger in size as the uptake in the left kidney
which gradually became intense and bigger in size as the uptake in the left kidney came down in excretion phase. The activity persisted even at the 3½ hours post-lasix image. The GFR calculated by the Gates’ protocol was found to be 89.4 ml/minute and was contributed by the left kidney only. With keeping the IVU and MRI findings in background, the appearance of activity at the lower part of left kidney could be explained which was in the hugely dilated collecting system and ureter. 99mTc-DMSA renal scan (Fig. 6) showed normal tracer concentration in left kidney and non-functioning right kidney consistent with IVU and MRI findings.

Fig. 2 : Delayed film of IVU showing no evidence of contrast excretion on right side and grossly dilated tortuous opacified left ureter. Also note the sacral agenesis and scoliosis of the lumbar spine with convexity to left with associated segmentation anomalies of the vertebral bodies

Fig. 3 : T1 weighted sagittal MRI image of the lumbosacral spine shows complete sacral agenesis. There is evidence of low lying spinal cord with a focal syrinx in it

Fig. 4 : T1 weighted coronal MRI image of the lumbosacral spine reveals focal syrinx in low lying spinal cord. Also note evidence of right renal agenesis and hypertrophied left kidney with corticomedullary differentiation maintained

Fig. 5 : 99mTc DTPA renogram showing prompt perfusion and good cortical uptake in the left kidney and absent perfusion and cortical uptake on the right side. From the sixth minute image onwards, another area of radioactivity seen just below left kidney which gradually became intense and bigger in size as the uptake in left kidney came down in the excretion phase
Caudal dysplasia sequence is a rare congenital disorder, characterised by the absence of the sacrum and defects of variable portions of lumbar spine, associated with anomalies from different systems. The terms caudal regression syndrome, congenital sacral agenesis, sacral regression are used synonymously to designate the same disorder. The prevalence of the disorder is estimated to be around 0.1-0.25 per 10,000 in normal pregnancies or around 1 in 60,000 births and 200-250 times higher in diabetic pregnancies. The etiology of the disorder is unknown, with maternal diabetes, genetic predisposition and vascular hypoperfusion have been suggested as possible causative factors. Sixteen percent of the affected are associated with maternal diabetes. Other proposed factors are hyperketonemia, hypoglycemia, somatomedin inhibitor excess and excess free oxygen radicals. CDS is probably due to a disturbance of the caudal mesoderm before the fourth week of gestation. Faulty retrogressive differentiation results in disruption of the maturation of the caudal portion of the spinal cord complex leading to varying degree of motricity deficits and neurologic impairment. Associated maldevelopment of the notochord results in various vertebral anomalies.

A wide range of abnormalities may occur including partial absence of the tailbone end of the spine causing no apparent symptoms, to extensive abnormalities of the lower vertebrae, pelvis, and spine. Anomalies of the central nervous, musculoskeletal, genitourinary, cardiac, respiratory and gastrointestinal systems are found frequently in association with caudal regression syndrome. They are enlisted in Table 1.

In this case syringomyelia was seen in association with CRS; this has not been described previously and needs inclusion in the list of associated anomalies.

The sonographic features in the intrauterine period are variable depending on the extent and severity of the defect. First trimester diagnosis is hard to accomplish because of the incomplete ossification of the sacrum at that time. A short crown-rump length and abnormal appearance of the yolk sac have been proposed as early sonographic signs of caudal regression syndrome. CDS can be detected in a fetus during the second trimester of pregnancy by ultrasound. The most typical findings are the absence of a few vertebrae, the shield-like appearance of the fused or approximated iliac wings and the decreased interspace between the femoral heads.

If detected early, pregnancy termination can be offered. Standard prenatal care is not altered if continuation of pregnancy is opted for. If born alive, the main goals of treatment include maintaining and improving renal, cardiac, pulmonary and GI function, preventing renal infection, and achieving continence. Extensive surgery in tertiary centre is usually needed to repair the defects. Orthopedic devices are useful for problems of the hip, back and legs. Features e.g. an imperforate anus, hydrocephaly, cleft plate or lip, and extra fingers or toes can be improved by corrective surgery. Physical therapy helps in preventing secondary deformities, skin ulcers, and achieving routine functions to improve the quality of life.

Sirenomelia, amniotic band syndrome, villus sampling limb defect, Kilippel Feil syndrome, Goldenhar syndrome are some...
of the diseases which should be considered in differential diagnosis of CDS. Knowledge of the characteristic feature of the above disorders helps easily to distinguish them from CDS. Till recently, sirenomelia or Mermaid syndrome was thought to be the most severe form of caudal regression syndrome. Fusion of the lower extremities is a typical finding of sirenomelia. Today its etiology is considered to be unrelated to CRS, embryonic vascular disruption postulated to be the responsible factor. Stevenson et al proposed the “vitelline artery steal” theory and inferred that sirenomelia resulted from a “vitelline artery steal”, which diverted blood and nutrients away from the caudal portion of the embryo to the placenta.

REFERENCES

Announcement

The Association of Physicians of India
Tirunelveli Chapter 2003-04

Inaugural function of API Tirunelveli Chapter was conducted at Raja Towers, Palayamkottai, Tirunelveli - 627002, on 25.5.2003.

Chairperson : JR Edwin
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