Case Reports

Sacral and coccygeal agenesis (Caudal Regression Syndrome)

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Abstract A 13 year-old Saudi female patient who has agenesis of lower sacral and coccygeal bones (Caudal Regression Syndrome) is herein reported. Clinical, radiological and MRI aspects are discussed and compared with other relevant publications.

This report has been written in order to provide new and unusual imaging of a relatively rare disorder.

Keywords: Sacrum and coccyx agenesis; Radiology MRI.

A 13 year-old Saudi girl was admitted to the Armed Forces Hospital, Southern Region, Kingdom of Saudi Arabia with increased frequency of micturition, repeated vomiting and pruritus.

On examination she was afebrile, in a stable state and with a normal blood pressure and pulse. She was not anemic but severe scratch marks were noted all over her body. The respiratory system was normal.

The axial skeleton was poorly developed with spinal curvature and a narrow pelvis. Equinovarus deformity and spasticity of lower limbs was noted. S1 and S2 reflexes were absent. Laboratory examinations were within normal limits apart from proteinuria and pyuria due to urinary tract infection.

Radiological examination The chest and skull radiographs were within normal limits. The thoracic spine was normal. Lumbar spine and sacral radiographs showed the presence of S1, S2 vertebral bodies, but the absence of lower segments of sacrum and coccyx.

Ultrasonography revealed bilateral hydronephrosis with reduction of renal parenchyma and hydroureters.

A cystogram showed that the urinary bladder was abnormally small, thick walled, spastic and with a trabeculated wall which could accommodate only a small volume of contrast. There was vesicourethral reflux on both sides with distention of the ureters and pelvicalyceal systems (Fig.1).

During the examination it was noted that the sacrum was abnormal and the distal segments were absent and that the bony pelvis was narrowed from side-to-side (Fig.2). Spasticity of lower limbs was noted. The above findings correspond well with the characteristics of the Caudal Regression Syndrome (sacral agenesis). Apparently in these disorders there is also atrophy of the cauda equina, (and possibly even of the caudal part of the cord).

For this reason it was deemed appropriate to perform magnetic resonance imaging (MRI) of the thoracic and lumbar spine as well as the pelvis. It was appreciated that surgical intervention was not likely be an option due to the character of the complex neurological malformation.

However, the patient had undergone long-standing urological investigation, and management which was modified by local factors. Eventually surgical intervention to augment the bladder capacity was consented to. The patient was able to remain dry for short periods. A regular stool softener was recommended and a daily bowel evacuation was advocated to maintain the quality of life of this girl.

During her most recent readmission for urosepsis, the opportunity was taken to obtain MRI examination of thoracic, lumbar and sacral spine as well as pelvis.

The results of MRI imaging The conus medullaris was located at the level T12 (unexpectedly high for children of this age (Fig.3). The cauda equina was small and somewhat atrophic. Agenesis of S3, S4, S5

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S5, the coccyx and the related nerve roots were noted. (Fig. 4). No evidence of sacral dysraphism with associated tethering of the cord and intraspinal lipoma was observed.

The small urinary bladder and distended ureters as well as the hydronephrotic changes in the pelvicalyceal systems bilaterally were also well seen (Fig. 5).

**Discussion**

Caudal Regression Syndrome is represented by a group of abnormalities ranging from lower lumbar to sacral spinal agenesis or hypoplasia. In those cases with agenesis, underdevelopment of the lower extremities can be observed, even in extreme conditions such as sirenomelia or mermaid deformity. Fused ilia or an iliac amphiarthrosis may also occur.

In those with sacral agenesis, as compared with sacral hypoplasia, neurologic deficit is more pronounced and is associated with clubfoot deformity, wasting of the muscles of the lower extremities, an incompetence of the urinary and anal sphincters. The neurologic deficit in these cases results from underdevelopment of the corresponding nerve roots and distal spinal cord. In sacral agenesis, intraspinal lipomas may be observed and these children frequently have
Fig. 3 - Mid sagittal T1-weighted image (TR=500, TE=11) of dorso-lumbar junction to demonstrate the conus at the D12 level.

Fig. 4a - Sacral agenesis: Mid sagittal T2-weighted image (TR=2000, TE=30) to show absence of sacrum below the S2 segment. Note the disordered nerve roots, dorsal and distal to the sacral remnant.

Fig. 4b - Sacral agenesis: Axial T1-weighted image (TR=500, TE=11). The level is S2. Note the absence of the dorsal bony arch which is replaced by a fibrous band. The cauda equina remnants are seen in the spinal canal.
cutaneous abnormality such as tufts of hair, dimples and subcutaneous lipomas as seen in spinal dysraphism.

In our case lower limb abnormalities (spasticity, wasting of muscle, clubfeet deformity) anal sphincteric and urinary tract problems were present. Chronic infection of the urinary tract was present.

According to our own views, as well as the literature, it is important to examine the sacrum in all patients initially referred for recurrent urinary tract infections. More obvious sacral abnormalities should be noted on simple clinical examination.

Many similar patients are born of diabetic mothers, but in our case this was not confirmed. No familial history was recorded in the family.

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References

الخلاصة:
تقرير عن مريضة سعودية تبلغ من العمر 13 عاماً تعاني من نقص النمو في العظام العجزية والعصعصية (متلازمة كودال الانتدابية). وقد ناقشنا في هذا البحث نتائج الفحص بالأشعة والتصوير بالرنين المغناطيسي وقارناها بالابحاث الأخرى التي نشرت في هذا المجال.

وقد قمنا بإعداد هذا التقرير بغية توفير صورة جديدة وغير معتادة عن مرض نادر.

الكلمات الرئيسية: نقص نمو العظام العجزية والعصعصية، الأشعة والتصوير بالرنين المغناطيسي.