Case Report

Atypical caudal regression syndrome with agenesis of lumbar spine and presence of sacrum – case report and literature review

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Context: Caudal regression syndrome is a rare disorder, not well described in the literature.

Findings: Authors treated two patients with congenital absence of thoracolumbar vertebrae and lower limbs paraplegia. Patients had hypoplasia of the lower trunk and extremities with motion between upper and lower torso. Imaging showed caudal spine agenesis, but cleft sacrum was present. Due to severe kyphotic deformity and spinal instability, deformity correction and posterior fusion was performed at the age 6 and 8. Finally, fusion was achieved in one case and stable but non-fusion kyphotic posture was observed in second.

Conclusion: surgery in caudal regression syndrome is challenging and bears high risk of complications.

Keywords: Caudal regression syndrome, Congenital spinal defect, Lower limb paraplegia, Spinal agenesis, Spine surgery

Introduction

Malformations of spine can be caused by abnormalities in cellular differentiation at the early stage of embryofetal development and they lead to either absence or defective structure of respective anatomical structures. The etiology of these deformations remains unknown, although such severe defects are usually induced by harmful factors operating at embryonic period.¹ Genetically determined spinal deformities are extremely rare.^{2,3} Congenital absence of spine / vertebrae is among the most infrequent groups of congenital defects. Most often reported are partial and total sacral agenesis, with the incidence below 0.5%.⁴ Currently, absence of a sacrum and partial absence of one or more vertebrae cephalad is classified as caudal regression syndrome.⁵ Despite great variety of defects in this syndrome, spinal malformations are always accompanied by anomalies of the viscera, especially of urogenital system.^{4,5} It is often associated with neurogenic bladder, which may lead to increased risk of renal impairment caused by recurring urinary tract

infections and urinary retention.^{6,7} Neurological deficits of various severity are often present.

The study presents two patients with congenital absence of thoracolumbar vertebrae with presence of hypoplastic sacrum.

Case presentation

In years 2001–2015 the authors treated two unrelated patients with congenital absence of thoracolumbar vertebrae, a female born 2005, and a male born 1995. They were born at 38 weeks with low birth weight, myelomeningocele was operated on in both patients within 24 hours from birth. The female patient was additionally diagnosed with hydrocephaly and had a ventriculoperitoneal shunt placed.

On admission, hypotrophic body composition and hypoplasia of the lower trunk and lower extremities was noticeable. Patients suffered paraplegia, lower limbs were in a “buddha” position with severe flexor contractures and spinal instability, with visible motion between upper and lower torso. Both patients had neurogenic bladder. They were wheelchair users, but they were also able to move on their hands.

Patient 1

The female, first presented to the Orthopedic Clinic at the age of 6. Physical examination revealed a typical

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clinical picture, consistent with the description above. Knee-joint contractures exceeded 90 degrees, with popliteal webbing present. A sharp-angle kyphotic deformity could be observed in the sitting position, significantly correctable on the prone extension test.

Radiographic imaging (X-ray, CT, MRI) showed block vertebrae of Th6 and Th7, agenesis of Th12 vertebral body, and total absence of lumbar spine (Fig. 1 and 2). The sacrum was present, but cleft. Dural sac and spinal cord were within spinal canal to Th8 level. In the last part of the dural sac neuromas were found. A cleft dural sac was visible caudally to the last thoracic vertebra.

The patient was scheduled for surgery due to severe kyphotic deformity and instability. At the age of 8 a posterior thoraco-sacral-pelvic instrumented fusion was performed, with an additional autograft acquired from fibula. Surgery was performed from posterior approach, deformity correction and fusion at the apex of deformity was done. Stabilization was done with use of hybrid construct: bilateral pelvis screws, two rods and sublaminar wires in thoracic spine.

Problems with wound healing caused by prominent hardware occurred in the postoperative period. Additionally, the patient suffered from recurring urinary tract infections (the patient was using incontinency pads). One year after the operation instrumentation was removed due to recurring infections of the wound. The last post-op checkup was conducted at the age of 11; the patient was using a wheelchair, and the neurological condition did not worsen. After removal of the instrumentation, correction was not maintained and the posture of the torso declined in the sitting position and was similar to the state prior to the operation. The shape of torso was with mild oblique kyphotic angulation, confirmed with X-ray. The spine behaved stable during examination, although there were no visible radiological signs of fusion at the apex of deformity.

Patient 2

The male, born in 1995, first presented to the Orthopedic Clinic at the age of 6 due to kyphoscoliotic spine deformation, with concomitant chest deformation and dislocation of both hip joints. Patient was able to sit independently when propped on forearms, with lower extremities extended in hip joints, crossed and flexed in knee joints, with popliteal webbing. Motion between upper and lower torso at the apex of the kyphotic deformation was visible during changing position. The patient suffered from rigid contractures and dislocations in hip and knee joints, bilateral clubfoot, was significantly hypotrophic and of low weight.

Radiographic, CT and MRI imaging revealed: only 9 vertebrae in the thoracic spine, total agenesis of the lower thoracic and upper lumbar spine, a sharp angular curve of the long axis of the spine. L4 vertebral body was dysplastic, whereas the L5 body and the sacral segment were cleft posteriorly. Cervical spine signals were normal, at Th4-Th5 level hydrosyringomyelic cavity was present. Below Th8 the spinal canal narrowed and terminated at the level of kyphotic curve of the spine (Fig. 3).

Inability to sit properly caused by extension contractures of the hip joints was most problematic for the patient. Therefore the treatment plan was divided into stages. In the first stage the proximal ends of femurs were resected. A few months later the patient underwent a spine surgery from posterior approach. The kyphotic gibbus was resected and stabilization with one rod and hooks was done. Following the surgery a cast brace was used. Scarcity of soft tissue in the spinal area as well as hardware prominence led to an infection of the wound and consequently necessitated the removal of the instrumentation. One year later, another surgical procedure was necessary due to big mobility at the level of deformity apex caused by spinal pseudoarthrosis. Anterior-posterior fusion was performed from posterior approach, followed by posterior stabilization with use of two rods placed through sacral foramina, two hooks and sublaminar wires in thoracic spine. After two years the instrumentation was removed due to hardware prominence. Fusion was preserved at two mobile levels of the spine. During the last follow-up checkup, at the age of 21, the patient was able to move independently on a wheelchair, and was studying. The hip joints and spine surgeries improved the quality of sitting through stabilization of the torso and flexion of the lower extremities in relation to it. The radiological imaging revealed angulation of the long axis of the spine at thoracolumbar junction, with no progression during last five years and spondylodesis visible at the apex of the deformation.

Discussion

Caudal regression syndrome (CRS) is a rare disorder, which in fact is a group of diverse congenital abnormalities, such as partial sacral agenesis, absence of a sacrum and part of lumbar spine.^{7–12} Genetic factors as well as factors related to the maternal diabetes mellitus have been reported.^{2,11–13} In the first CRS report Duhamel presented two most characteristic types of the disorder: mermaid with lower extremities fusion, and anchipodal type with lower limbs not fused but flexed in knee joints, abducted in hip joints, with popliteal webbing.⁹ Later, the term CRS was also applied to minor and major defects of a sacrum.^{8,11,12} The two patients presented

Figure 1 Patient 1. Lateral x-ray before surgery, after and at follow up.

in this paper correspond to the later type in Duhamel's classification; however, what draws attention is atypical presence of the sacrum and L5 vertebral body, and such extensive partial agenesis of lumbar spine. Typically, patients suffer from characteristic for this syndrome paraplegia caused by neurologic deficits, neurogenic bladder and recurring urinary tract infections which may lead to renal failure.^{4,8,14} Most authors describe patients just after birth or in neonatal period, and there are mostly case reports.^{9,13} Only few of them consider quality of life of these patients underlining the importance of stable sitting position without hand support.⁵ To achieve this goal, knee and hip joint contractures may require (or not) surgical intervention,

depending on the patient.¹⁵ Spino-pelvic instability is another problem causing declined torso position. Similar problem may occur in children with myelomeningocele, where kyphotic deformity may present "a functional impediment in regard to sitting".¹⁶ In these patients surgery is required to correct sagittal spine balance and achieve stable sitting position without hand support.¹⁶ Patients with caudal regression syndrome have similar problems with unstable trunk and kyphotic deformity, and this may be an indication for surgical treatment, although there are no long-term, big patients' series observations.^{5,12,17,18} Our patients were operated on due to spinal-pelvic instability and kyphotic curve. In the male patient it was possible to

Figure 2 Patient 1. CT scan with 3D reconstruction.

Figure 3 Patient 2. Lateral x-ray before surgery, after and at follow up.

achieve union, though unfortunately in a kyphotic position; in the female patient, however, the attempt to achieve stabilization in the same area has been unsuccessful. Obtaining spondylodesis in these patients is difficult. Also, it should be remembered, that regardless what method is used to stabilize the spine, in patients of such hypotrophic body build there is high risk of complications related to hardware prominence: abrasions and damages (lesions) to the skin immediately above the instrumentation and wound infections.^{5,18} We chose as low profile implants as possible in those times, however, hardware prominence was still main cause of treatment failures in our patients, as well. It is possible, that a two-stage primary procedure with the use of vascularized bone grafts, followed by multiple re-operations, as proposed by Ferland *et al.* is the most effective for this group of patients.¹⁸ A better result was achieved in the second patient, in which case a two-stage procedure plan was followed, where despite the necessity to re-operate and complications, it was possible to obtain stabilization of the spine and improve sitting position. We did not achieve stable correction of spinal deformity – may be new generations of implants will allow surgeons achieve stable fusion and correction without implant-skin problems in these group of patients. Surgery must of course be considered individually, since the clinical presentation is not uniform, and patients present also many underlying conditions. In the non-operative cases conservative management with chair modification may be the alternative.

Conclusions

Partial agenesis of thoracic and lumbar spine is a serious congenital defect causing total paralysis of lower extremities and spinal-pelvic instability. Successful surgical

treatment of spinal deformities in this group of patients is challenging and bears high risk of complications, mainly implant-related.

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