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Letter to the Editor

Clinical and Radiologic Characteristics of Caudal Regression Syndrome in a 3-Year-Old Boy: Lessons from Overlooked Plain Radiographs

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Conflict of Interest
The authors have no financial conflicts of interest.

ABSTRACT

Caudal regression syndrome (CRS) is a rare neural tube defect that affects the terminal spinal segment, manifesting as neurological deficits and structural anomalies in the lower body. We report a case of a 31-month-old boy presenting with constipation who had long been considered to have functional constipation but was finally confirmed to have CRS. Small, flat buttocks with bilateral buttock dimples and a short intergluteal cleft were identified on close examination. Plain radiographs of the abdomen, retrospectively reviewed, revealed the absence of the distal sacrum and the coccyx. During the 5-year follow-up period, we could find his long-term clinical course showing bowel and bladder dysfunction without progressive neurologic deficits. We present this case to highlight the fact that a precise physical examination, along with a close evaluation of plain radiographs encompassing the sacrum, is necessary with a strong suspicion of spinal dysraphism when confronting a child with chronic constipation despite the absence of neurologic deficits or gross structural anomalies.

Keywords: Constipation; Sacrum; Neural tube defects; Congenital abnormalities; Abdominal radiography

INTRODUCTION

Childhood constipation is a common symptom encountered by general pediatricians as well as pediatric gastroenterologists. More than 90% of children with constipation are diagnosed with functional constipation without identifying specific organic causes [1].

Caudal regression syndrome (CRS), also referred to as sacral agenesis syndrome, caudal dysplasia or caudal agenesis, is a rare congenital malformation caused by a developmental failure in neuralization during the early stages of gestation [2]. It is characterized by variable levels of spinal agenesis, accompanied by caudal vertebral malformation and structural anomalies in the musculoskeletal, genitourinary, and gastrointestinal (GI) systems [3]. Most cases are diagnosed at birth or during a prenatal examination by noting an abnormal appearance in the pelvis and lower extremities of the newborn or fetus [4,5].

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We describe a toddler with intractable constipation from infancy who we diagnosed with CRS and present his long-term clinical course.

This case series was approved by the Institutional Review Board (IRB) of Kangwon National University Hospital (IRB No. 2020-05-013) and the requirement for informed consents was waived due to the retrospectively collected data.

CASE REPORT

A 31 month-old boy was referred to the pediatric GI department for evaluation of intractable constipation. He had difficulty defecating with a frequency of 2 bowel movements per week requiring intermittent enemas and laxatives from the time that he was 2 months of age. His mother reported that his symptom was difficult to manage, because even a small amount of laxative often caused diarrhea.

He was born to a diabetic mother at the gestational age of 37 weeks with a birth weight of 2.65 kg. The mother stated that her blood glucose had been difficult to control with insulin since the early weeks of her pregnancy. At 11 days of age, he had a history of admission for hypocalcemic tetany without any identified underlying disease. He was diagnosed with a congenital hearing impairment through the newborn hearing screening test. He had also been hospitalized several times for febrile illnesses, such as influenza infection, bronchiolitis, and viral pneumonia.

On examination, his height was 84 cm (10th–25th percentile), and his body weight was 12 kg (10th–25th percentile). He was active and had low-set ears wearing hearing aids. His abdomen was slightly distended, with hard stools noted on a digital rectal examination. In addition, we observed that he had small, flat buttocks with bilateral buttock dimples and a short intergluteal cleft (**Fig. 1**). He passed normal mental developmental milestones except for a delay in language development, and there were no motor deficits or gross deformities in his extremities. His mother reported that he showed urinary dribbling and a weak urine stream since infancy.

When we reviewed the plain radiographs, including those taken during previous hospitalizations, no abnormal findings were initially recognized, except for an excessive amount of stool from the colon to the rectum (**Fig. 2A-D**). Under the suspicion of spinal

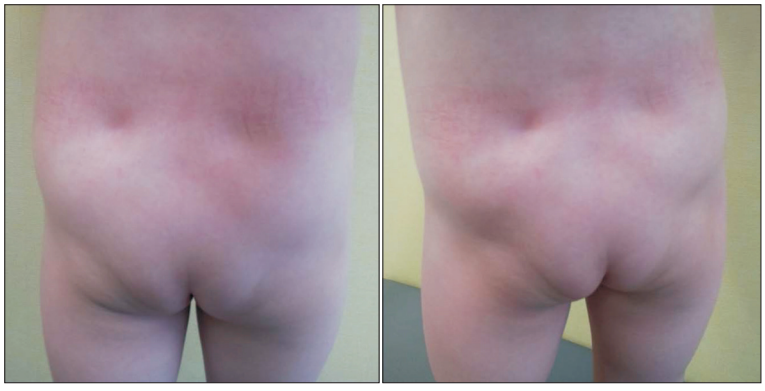

Fig. 1. Posterior view of the pelvis of the patient in the standing position. He showed a short intergluteal cleft, flattened buttocks, bilateral buttock dimples, and small gluteal masses.

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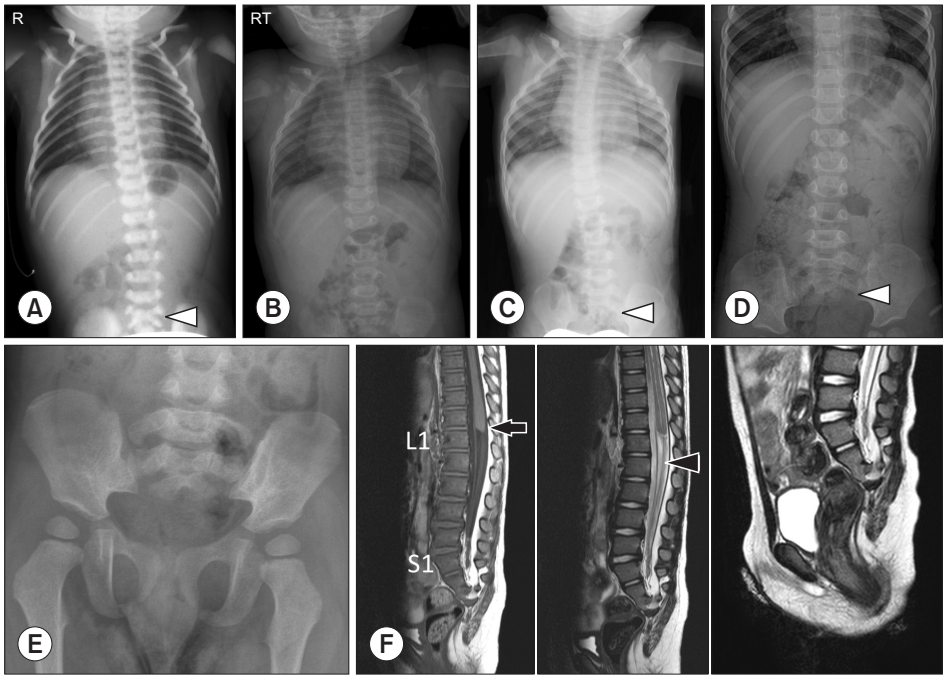

Fig. 2. Serial plain radiographs of the abdomen and sagittal magnetic resonance imaging of the spine. A retrospective review of abdominal X-rays performed during admission at the age of 10 days (A), 4 months (B), 10 months (C), and 31 months (D) demonstrated subtotal sacral agenesis (white arrowhead). Pelvic X-ray (E) after disimpaction revealed the absence of the distal sacrum and the coccyx. Magnetic resonance image of the spine (F) showed a club-shaped conus medullaris, which abruptly terminates at the level of T12-L1 (black arrow) and a thickened filum terminale (black arrowhead). It also demonstrated the absence of the distal sacrum and the coccyx.

dysraphism, however, we performed another pelvic X-ray, after disimpaction with an enema, that finally revealed the absence of the distal sacrum and the coccyx (**Fig. 2E**). Magnetic resonance imaging (MRI) of the spine demonstrated an abrupt termination of the conus medullaris at the T12 to L1 level and a thickened filum terminale (**Fig. 2F**). A voiding cystourethrogram with contrast showed poor filling in the bladder, suggesting a neurogenic bladder. No other remarkable findings were identified through abdominal and kidney ultrasonography, echocardiography, barium enema, and brain MRI.

After being diagnosed with CRS, he was followed up over a 5-year period in the department of Pediatric GI, Urology, Neurosurgery, Rehabilitation, and Otorhinolaryngology. He has been wearing diapers for his urinary incontinence and has continued to use laxatives or enemas whenever he has difficulty with bowel movements. He did not have other progressive neurological deficits that need other interventions, except for a mild delay in language and intellectual functioning.

DISCUSSION

We described the case of a boy with subtotal sacral agenesis, accompanied by bowel and bladder dysfunction, who was confirmed to have CRS. His only clinically significant problem, defecation difficulty, had long been considered functional constipation. His small and flat buttocks, which are characteristic features of sacral agenesis, and his urinary incontinence were difficult to detect during the neonatal-infant period when infants spend most of their time wearing diapers and lying down.

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Table 1. Clinical characteristics of cases with caudal regression syndrome published since 2000

Cases	Year of report	Age at diagnosis	Sex	Maternal diabetes	Symptoms at presentation	Combined anomalies	Outcome
Aslan et al. [4]	2001	22 weeks of GA	M	NR	Prenatal diagnosis	Absent lumbosacral vertebra, hypoplastic pelvis, missing ribs, flexion contractures of lower extremities, club feet, ventricular septal defect	Termination of pregnancy
Zaw and Stone [7]	2002	At birth	F	Yes	Gross anomalies detected at birth	Absent lumbosacral vertebra below L4, hypoplastic low extremities	Delivered at 33 weeks of GA
Singh et al. [6]	2005	2.5 years of age	M	No	Gross anomalies detected at birth	Imperforated anus, lower limbs deformities, total sacral agenesis, partial lumbar spinal agenesis, undescended left testis, absent left kidney	Colostomy, orchiopexy
Shah et al. [8]	2006	3 years of age	F	No	Urinary and bowel incontinence, difficulties to walk	Sacral agenesis (below S1), terminal cord syrinx, left-sided sacral rib	
Krenova et al. [9]	2010	22 weeks of GA	F	Yes	Prenatal diagnosis	Pelvic bones agenesis except for four segments of sacral bone, aplasia of both femoral bones and fibulas, hypoplastic both tibial bones, micrognathia, palatoschisis, hypoplastic left eye ball, both ears deformities	Delivered at 35 weeks of GA
Kokrdova [5]	2013	18 weeks of GA	M	Yes	Prenatal diagnosis	Arnold-Chiari malformation, lumbar myelocele, absent sacrum, hypoplastic lower extremities, feet deformities, hypoplastic kidneys	Termination of pregnancy
		20 weeks of GA	F	Yes	Prenatal diagnosis	Deformities of lower limbs and feet, shortening of lower spine, absent left kidney, truncus arteriosus communis	Termination of pregnancy
Sharma et al. [10]	2013	10 years of age	F		Lower extremities weakness, urinary incontinence	Sacral agenesis (below S2), syringohydromelia, anterior epidural lipoma, anterior lipomyelocytocele, bilateral hydronephrosis, bilateral hydroureter	Surgery for neurogenic bladder at 5 years old
Duncan et al. [11]	2014	18 weeks of GA	F	Yes	Prenatal diagnosis	Total sacral agenesis, hypoplastic iliac bones, shortening of lower extremities, club feet, hypoplastic kidneys	Delivered at 40 weeks of GA

GA: gestational age, M: male, F: female, NR: nor reported.

CRS is a neural tube defect affecting the terminal spinal segment. The clinical features and physical findings of CRS are variable, depending on the extent of the defects. It causes extensive neurological deficits in the lower limb, as well as loss of bladder, renal, and bowel function that present as fecal and urinary incontinence [3]. It can be a progressive neurological disorder if it is associated with a tethered cord. Associated anomalies include pelvic deformity, femoral hypoplasia, renal agenesis or dysplasia, genital duct anomalies, such as hypospadias or bicornuate uterus, or anorectal malformation as imperforated anus [6]. Although the etiology of CRS is not clearly defined, maternal diabetes is considered to play a critical role. Genetic factors, vascular hypoperfusion, and some teratogenic drugs have also been suggested as possible causative factors [6]. The clinical characteristics of published cases since 2000 have been reviewed and presented in **Table 1** [4-11].

In this case, our patient did not show any gross deformities, except for flat and small buttocks. Because his first and second sacral bones (S1 and S2) were present and the iliosacral articulation was preserved, there was no spinopelvic instability or lower extremity malformation. According to the radiologic classification of sacral agenesis, it corresponds to type III, the most common type, where the transverse pelvic diameter is relatively maintained at the point where the preserved S1 articulates with the ilia [2]. His low-set ears and congenital sensory-neural hearing loss have not yet been reported as the associated anomalies of CRS.

Approximately 3% of children with intractable constipation have been found to have a lumbosacral spine abnormality, even without the presence of neurologic symptoms [12]. According to expert opinions, history taking and physical examination are sufficient to

diagnose functional constipation; hence, any testing including abdominal radiography is not recommended before initiating treatment in the absence of alarm signs and symptoms suggesting the presence of underlying causes [13]. In clinical settings dealing with children, plain radiography is a widely available, inexpensive, and easily obtained imaging test. Moreover, it is a good diagnostic tool to check for abnormalities in the skeletal system. Feces in usual radiographic sites of the sacrum often make the evaluation of the distal lumbar, sacral, and coccygeal portions challenging; however, a careful examination of abdominal radiographs focusing on the sacrum can provide definitive clues when spinal dysraphism is suspected [14]. For our patient, the plain radiographs performed at 11 days of age showed that the ossification centers of S1 and S2 were in an abnormal position. In addition, we recognized that there were only two visualized sacral bones in the X-rays performed even at the 10 months of age.

We present this case to highlight the fact that spinal dysraphism including CRS should be kept in mind for a differential diagnosis when examining a child who suffers from chronic constipation refractory to standard medical treatment despite the absence of neurologic deficits or gross structural anomalies. In addition, a close evaluation of radiographs encompassing the sacrum is necessary not to miss the spinal dysraphism, along with an accurate physical examination.

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