Clinical Case Report

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Urine retention as the first presentation of congenital absence of the sacrum

A case report of a rare clinical phenomenon

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Abstract

Rationale: Congenital absence of either a part of or the entire sacrum is rare, and only a few such cases have been reported until now. Most cases showed vertebral, pelvic, neurological, and visceral abnormalities, and the cases were usually quite serious. Only in rare cases was there urine retention without other symptoms.

Patient concerns: We describe the case of a 28-years-old man who presented with urine retention.

Diagnoses: Urodynamic study confirmed that he had urinary retention, and magnetic resonance imaging (MRI) showed the absence of the sacrum.

Interventions: He was treated with electro-acupuncture and medications to improve nerve function.

Outcomes: His symptoms showed clear improvement.

Lessons: Although this case is rare, it indicates that the spine and pelvis may need to be examined when unexplained urinary retention occurs.

Abbreviations: CT = computed tomography, MRI = magnetic resonance imaging.

Keywords: congenital defect, sacrum, sacrum dysplasia, urine retention

1. Introduction

Congenital absence of the sacrum is rare, and only a few such cases have been reported until now. The reported incidences rate of 0.01 to 0.05 cases per 1000 live births,[1] and the etiology is still not clear. Sacral agenesis has a wide spectrum of phenotypes,[2] and most cases showed vertebral, pelvic, neurological, and visceral abnormalities. We now depict a case of a young man who was congenital absence of the sacrum and only presented with urine retention.

1.1. Case

A 28-year-old man who had long-term frequent micturition and urgent urination was admitted to the Department of Nephrology of our hospital. He was a full-term baby and his parents had no similar symptoms. There were no abnormal signs upon physical examination. Laboratory studies, including routine blood evaluation, renal function, electrolyte, and routine urine tests were normal. Computed tomography (CT) and ultrasonography of the abdomen revealed hydronephrosis and large amounts of urine retention in the bladder (Fig. 1). A urodynamic study showed detrusor underactivity, considering a neurogenic bladder. He was admitted to the Department of Orthopedic Surgery, and physical examination revealed the absence of a coccyx. Magnetic resonance imaging (MRI) findings showed sacral canal thickening, an abnormal nerve and sacral canal, and sacrum dysplasia (Fig. 2). Genetic testing revealed the transcription factor Brachyury (T) gene had no mutations. From the imaging features, it was determined that urinary retention was caused by congenital absence of a section of the sacrum and sacral nerve, which led to bladder detrusor dysfunction. To improve the symptoms, indwelling urinary catheter was used to relieve urinary retention and neurotrophic drugs were prescribed. A recent study had shown that electroacupuncture was effective and safe in regulating nerve function andt sacral nerve stimulation can treat partial sacral agenesis for fecal incontinence,[3–6] Thus, acupuncture was performed. The acupuncture points are Sanyinjiao (SP6), Yinlinquan (SP9), Xingjian (ST37), and Zhaohai (KI6). After sterilizing the skin, filiform needles were inserted 3 to 8 cm into bilateral SP6, SP9, ST37, and KI6 vertically and slowly without any manipulation until they pierced the muscle layer. Then, the needles were twirled, lifted and thrusted. Each session last for 30 minutes per day, continuously, for 4 weeks. Finally, the symptoms significantly improved. The urodynamic studies showed

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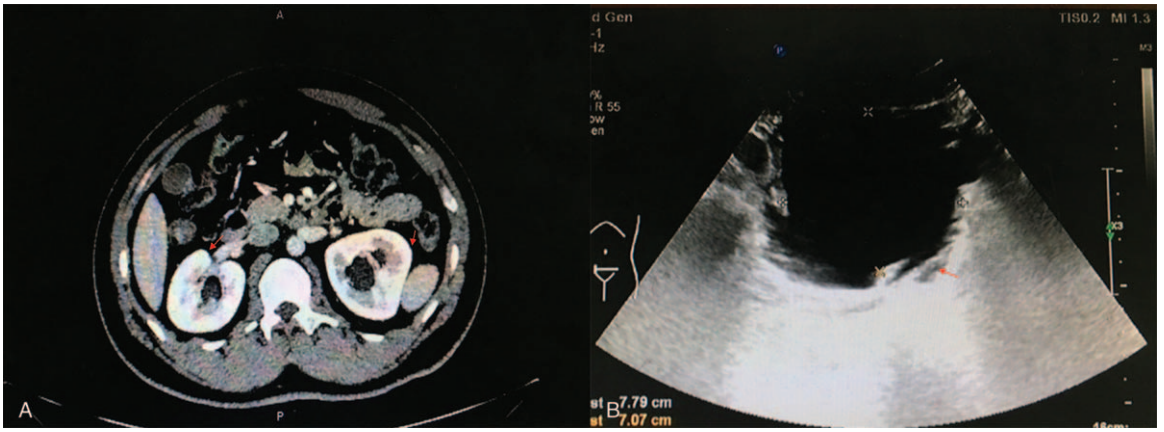

Figure 1. (A) CT scan showing hydronephrosis. (B) Ultrasonographic image showing urine retention in the bladder. CT = computed tomography.

urodynamic parameters improved in first sensation at filling, detrusor pressure at filling, and bladder compliance at filling. and continuous follow-up will be performed the next 1 year.

This study complied with the Declaration of Helsinki and was approved by the Human Ethics and Research Ethics Committees of the Fourth Hospital of Hebei Medical University. The participating patient provided written informed consent.

2. Discussion

Congenital absence of the sacrum is rare, and the etiology is still not clear. Some studies have shown that exposure of the embryo to fat solvents, high temperatures, and lithium salts and deficiency of vitamin A may cause congenital absence of the sacrum in animal.[7,8] Another study has shown that human T gene and its exclusion as a major candidate gene for sacral agenesis in familial forms and maternal diabetes may cause sacral agenesis in sporadic case.[2,9] One study revealed that sacral dysgenesis was associated with terminal deletion of chromosome 7q.[10,11] The current study showed copy number variation deletions in genes, which may be the etiology of sacral agenesis.[12]

Clinical features of congenital absence of the sacrum range from simple sacral abnormalities to sacral agenesis with accompanying musculoskeletal, visceral, and neurological ab-

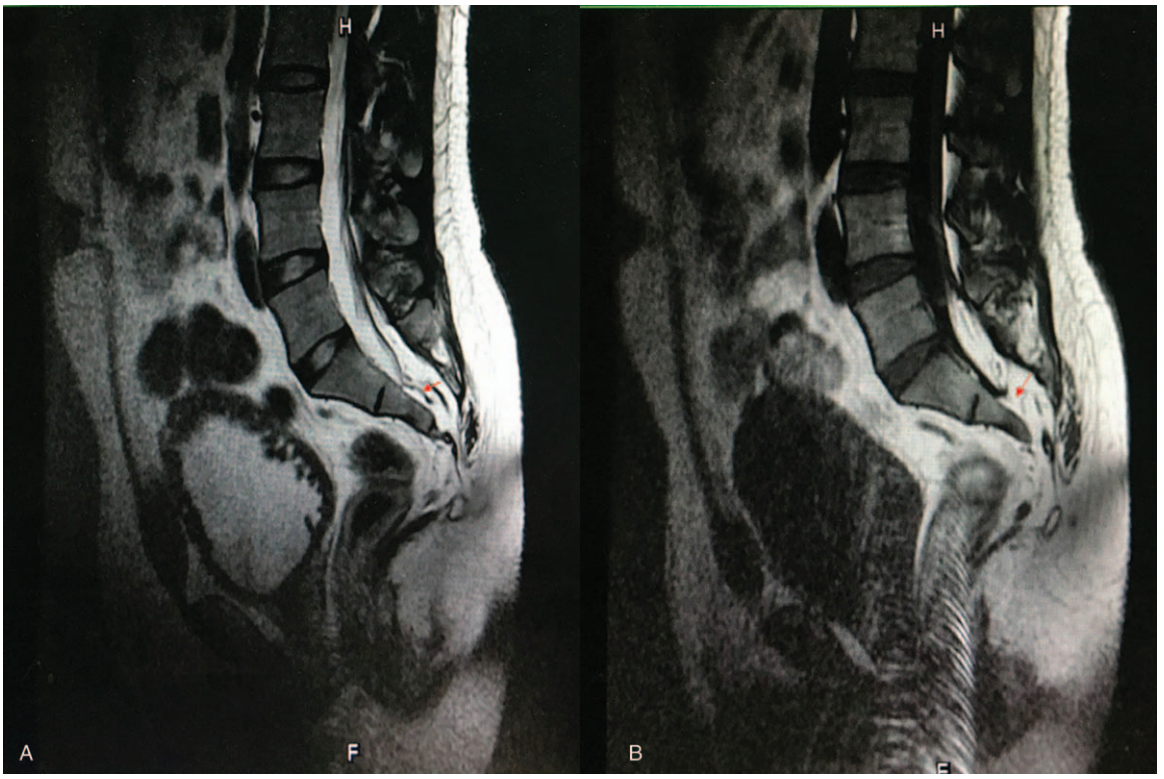

Figure 2. (A) T2-weighted MRI and (B) T1-weighted MRI showing second sacral dysplasia and the absence of the remaining lower sacrum and coccyx. MRI = magnetic resonance imaging.

normalities.[2] The visceral abnormalities include urogenital, cardiovascular, and pulmonary anomalies, for example, anal atresia, hermaphroditism, and hypospadias. The neurological abnormalities may include urine and fecal incontinence, muscles atrophy, and sensory changes.[7,13] There are several classifications for sacral agenesis.[14,15] Imaging examinations can result in a confirmed diagnosis of sacral agenesis, which is often associated with anomalies of multiple organs and may severely affect a patient's quality of life. Therefore, early diagnosis and treatment are necessary.[16] Sacral agenesis can be identified through prenatal ultrasound screening, and early screening should be conducted for fetuses at a high risk for anomalies.[16,17] The main treatment for sacral agenesis is correction of deformities and orthopaedic intervention.[14,18,19] Moreover for neurogenic bladder, the current treatment methods include medications, neuromodulation, botulinum toxin A injection, surgical procedures, issue engineering, stem cell transplantation, and gene therapy, besides, Traditional Chinese Medicine such as acupuncture. A review demonstrated that acupuncture could have a beneficial effect on neurological function recovery.[6,20] For this patient, only presented with neurogenic bladder without other symptom, we will continue follow-up with clinical examination, renal laboratory tests, imaging surveillance of the upper urinary tract, urodynamic study, and cystoscopy for at least 1 year.[21] So far, we have followed-up of 6 months.

3. Conclusion

The patient had repeatedly hospitalized for urinary retention and was diagnosed as neurogenic bladder, but no one found sacral agenesis until admitted to our hospital. Although this case is rare, it indicates that the spine and pelvis may need to be examined when unexplained urinary retention occurs.

Author contributions

Conceptualization: Hongzeng Wu, Peng Qi, Helin Feng.
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