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Case Report

Sacral Agenesis and Secondary Complications of Neurogenic Lower Urinary Tract Dysfunction

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Abstract

Objectives: To present a case of a patient with sacral agenesis and chronic renal failure in whom enterocystoplasty was used to treat vesico-ureteral reflux and impaired storage function of neurogenic bladder.

Methods: To describe the clinical characteristics of an adult patient with sacral agenesis, who presented at emergency department with urinary fistula on the left flank and sepsis syndrome. Also, to describe the diagnostic and surgical management and outcome over five years of follow-up. The therapy is focused on the bladder to achieve the primary goals of maintaining residual renal function and attaining urinary continence. It was primary outcome measure.

Results: Enterocystoplasty was performed in an attempt to prevent further upper tract deterioration and to treat urinary incontinence. Postoperative retrograde cystography with straining showed absence of vesico-ureteral reflux and neo-vesica with significant capacity. Clean intermittent catheterization was recommended. After five years of close follow-up monitoring the patient presented slowing deterioration of renal failure and good continence during the day. The patient was satisfied with her quality of life. The only remaining problem was sporadic incontinence over the night.

Conclusion: Neurogenic lower urinary tract dysfunction is a constitutional part of variable clinical features of this rare congenital anomaly. Prompt diagnosis and surgical treatment of a vesico-ureteral reflux is advised to prevent damage to upper urinary tract. Enterocystoplasty remains an important option to reverse upper tract deterioration, and to treat socially unacceptable incontinence. This case strongly suggests the need for early diagnosis and long-term periodic follow-up in sacral agenesis.

Keywords: Chornic; Renal; Failure; Congenital; Anomaly; Foot; Deformity; Neurogenic; Lower; Urinary; Tract; Dysfunction; Sacral; Agenesis; Vesico-ureteral; Reflux

Introduction

Caudal regression syndrome, or sacral agenesis (or hypoplasia of the sacrum), is a rare birth defect. Sacral agenesis is a congenital lesion of the sacrum. It is described in a group of disorders where a portion of the caudal spine is absent [1]. Renshaw classified the patients according to the amount of sacrum remaining and the characteristics the articulation between the spine and the pelvis [1]. Type I is either total or partial unilateral agenesis of the sacrum. Type II is partial sacral agenesis with bilateral symmetrical defect, a normal or hypoplastic first sacral vertebra and stable articulation between the ilia and the first sacral vertebra. Type III is total sacral agenesis and the iliac bones articulate with the lowest available segment of the lumbar spine. Type IV is total sacral agenesis and the iliac bones are fused posteriorly along the midline.

This condition is rare, occurring in 0.005% -0.01% of the population. A higher frequency, in the range of 0.1%-0.2%, has been observed in children of diabetic mothers [2,3]. Approximately 16%-20% of children with sacral agenesis have mothers with diabetes mellitus [3]. It is thought that increased blood sugar levels and other metabolic problems related to diabetes may have a harmful effect on a developing fetus, increasing the likelihood of developing caudal regression syndrome. The risks to the fetus are further increased if the mother's diabetes is poorly managed. The specific etiology of sacral agenesis remains unknown. According to one hypothesis it could be teratogenically induced or spontaneous genetic mutation [1].

Disorders of the genitourinary tract in patients with sacral agenesis occur frequently, but neurogenic bladder is found the most common abnormality [1]. In our case the diagnosis was overlooked and the follow-up was poor. We report our experience of the treatment and long-term follow-up in a patient with sacral agenesis and chronic renal failure which was caused by overlooked neurogenic bladder.

Case Presentation

A 34-year old woman presented at emergency department with fewer, high temperature and abnormal leakage of urine on left flank (Fistula Urinaria). Clinical examination findings and laboratory tests results showed sepsis syndrome and renal failure. Ultrasonograhy revealed bilateral hydronephrosis without measurable renal parenchyma on the left kidney and hyperechogenic reduced thickness parenchyma on the right side. On the conservative treatment the patient recovered. Urinary fistula on the left flank remained.

The patient first experienced pronounced discomfort urinating at the age of nine. She underwent testing and an operation. A bilateral ureterocystoneostomy was carried out. After the operation, difficulties during urination persisted, varying in time and intensity. Conservative therapy did not yield adequate results. The patient was sporadically checked up afterwards.

She urinated in a sitting position, with straining. The urinary

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flow was intermittent. The patient was occasionally incontinent. Involuntary discharge of urine was meager, and was not connected with the change of the body position or with stressful situations. She did not have overwhelming drives to urinate. She was aware of the involuntary discharge of urine. The patient was continent feces wise. In the last year, the patient suffered frequent urinary infections accompanied with high temperature. Several days before the admission to hospital, she noticed the appearance of urine at the opening in the skin on the left flank.

Physical examination determined the position of the urinary fistula on the left flank, the hypotrophy of the muscles of the lower extremities and orthopedic deformity of both feet (Figure 1). Neurological examination registers preserved perineal sensation, preserved tonus of the external anal sphincter, and the ability of intentional constriction of the external anal sphincter. Laboratory testing registered creatinine clearance of 23 ml/min with serum creatinine of 296 mmol/l.

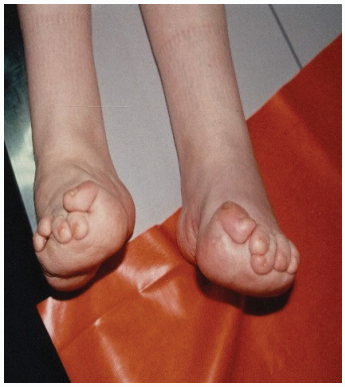

Figure 1: Foot deformity in the patient with sacral agenesia.

X ray examination showed agnesia of the sacrum and spina bifida occulta from L2 to L5 (Figure 2 and 3).

Retrograde cystography showed passive bilateral vesico-ureteral reflux gradus V and bladder of small capacity with thickening wall (Figure 3). Cystoscopy confirmed diagnosis of trabecular bladder with low filling volume. At the volume of 50 ml-70 ml, liquid leaks along the casing of the apparatus. We performed left nephrectomy. A month later, we performed a "clam" enterocystoplasty as well.

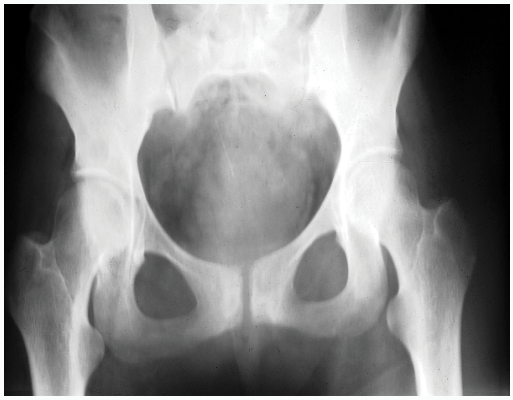

Figure 2: Anteroposterior roentgenogram - Type II sacral agenesis with bilateral symmetrical defect, a normal or hypoplastic first sacral vertebra and stable articulation between the ilia and the first sacral vertebra.

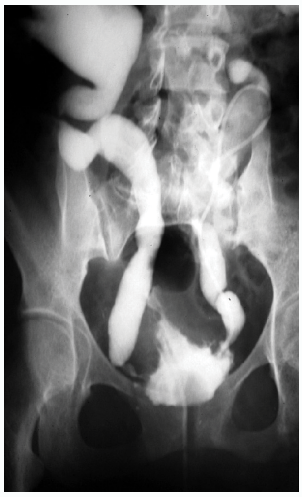

Figure 3: Retrograde cystography.

Results

Control retrograde cystography and cystography with strain showed the absence of vesico-ureteral reflux and bladder of significant capacity (Figure 4).

Clean intermittent catheterization was recommended since postvoided residual urine was over 500 ml. The patient was continent and satisfied with the quality of her life. The only insoluble problem remaining was nighttime incontinency. Involuntary emission of urine during sleep occurred without stimulation that would wake her up. The patient was checked at regular intervals.

Five years subsequent to augmentative cystoplastics, the value of clearance creatinine was 16 ml/min at the values of urea in the serum of 15.9 mmol/l and creatinine 406 μm mol /l. Echosonographic examination registered residual stasis of the remaining right kidney with hyperechogenic and reduced renal parenchyme.

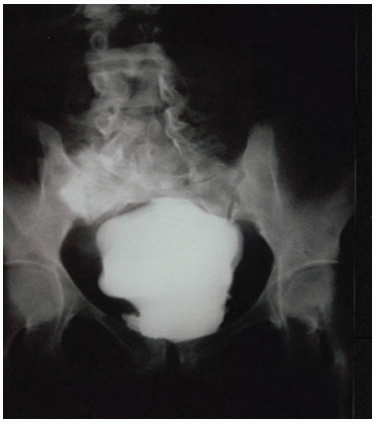

Figure 4: Retrograde cystography after augmentation enterocystoplasty.

Discussion

Sacral Agenesis (SA), also called caudal regression syndrome is a severe form of abnormal sacral development. Affected individuals have abnormalities of the musculoskeletal system most frequently involving lower extremity abnormalities (Figure 1).

Patients with SA have variable deficiencies in motor and sensory

function below the lowest level of the remaining normal spine [2]. There is also an increased frequency of spinal cord abnormalities in patients with sacral agenesis, including syrinx, tethered cord, lipoma, and lipomyelomeningocele [4]. Wilmshurst et al. [5] studied 22 children with sacral agenesis and associated neuropathic bladder retrospectively. Twenty-one children had abnormal neurology in the lower limbs. Videourodynamics showed neuropathic vesicourethral dysfunction in all children and vesicoureteric reflux in 10. However, patients do not necessarily present with neurological deficiency or external evidence of sacral agnesia. Symptoms and signs of neurogenic dysfunction of the bladder can be concealed, and so the diagnosis may be overlooked, delayed or suspended [6,7]. Although, SA almost invariably produces functional involvement of the bladder and recurrent urinary tract infection, the evolution may be benign [8]. The type and degree of neurogenic bladder may vary from one patient to the next [7]. Extensive examination must be undertaken to determine exact pathophysiology of lower urinary tract [5-9]. Some authors consider that magnetic resonance imaging screening should be ordered for patients with characteristic features or x-ray evidence of SA, anorectal, and genitourinary anomalies or other complex malformations of the caudal region [9].

Therefore, neurogenic lower urinary tract dysfunction constitutes a true disorder of neurophysiology of the lower urinary tract in patients with sacral agenesis. Detailed anamnesis including urinary symptoms is important. Most patients with SA had persistent dribbling of urine on presentation associated with frequency, urgency, recurrent urinary tract infections, failure to respond to medication, and/or constipation [5-7]. In this case, the disturbance of the symptoms of urination, and primarily incontinence pointed to the dysfunction of the lower portion of the urinary tract. Symptoms of urinary incontinency or persistent dribbling in patients with congenital anomalies of the spinal cord are significant, and the manifestation of vasico-ureteral dysfunction is obvious [10].

In this case, the patient came for urologic examination in the late stadium of the disease, presenting secondary complications NLUTD. Chronic renal insufficiency was pronounced. Clearing of creatine was 23 ml/min.

Vasico-ureteral reflux and frequent urinary infections were the root cause of the chronic renal insufficiency, pronounced hydronephrosis on both sides and the urinary fistula on the left kidney. The only therapeutic option was enterocystoplasty. It aimed at slowing down deterioration of renal failure and eliminating urinary incontinence.

Conclusion

Apart from congenital anomalies in urinary tract, secondary complications NLUTD may arise in sacral agnesia. NLUTD is a part of rare congenital anomaly of sacrum agnesia. Comorbidity of the urinary tract is significant and can be a life-threatening disease. Prompt diagnosis and surgical treatment of a vesico-ureteral reflux is advised to prevent damage to upper urinary tract. Enterocystoplasty remains an important option to reverse upper tract deterioration, and cure socially unacceptable incontinence. This case strongly suggested the need for early diagnosis and long-term periodic follow-up of patients with sacral agenesis.

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