DOI: 10.7860/JCDR/2021/49045.15165

Case Report

Surgery Section

Sacral Agenesis- Its Urological Impact and Management

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ABSTRACT

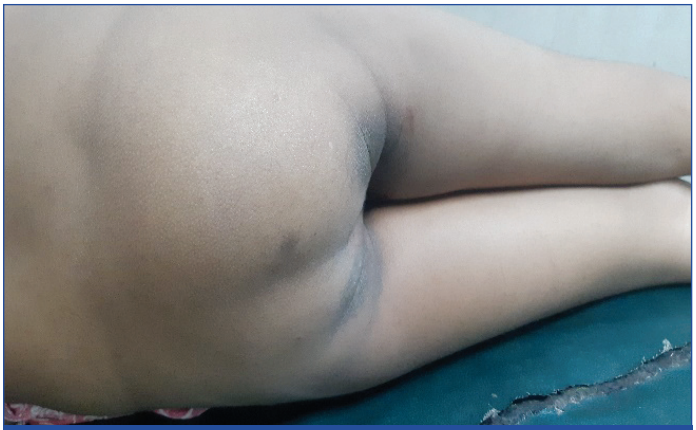
Sacral agenesis is a rare developmental disorder, in which there is absence of part or all of two or more sacral vertebral bodies. It is mostly associated with dysfunction of bowel, bladder and neuromuscular system of lower limbs. Voiding impairment (neurogenic bladder), Vescico-Ureteric Reflux (VUR) together with urinary tract infection contribute to renal injury. Here authors are reporting an 11-year-old female, who was diagnosed to have sacral agenesis with anorectal malformation at the age of six months, leading to anal transpositioning done at that age, she presented to the Urology Department with acute kidney injury. On evaluation, she was found to have overactive bladder with increased bladder capacity and bilateral grade 5 VUR. After recovery from acute kidney injury with conservative management and catherization, she was discharged with the advice of clean intermittent catheterization and anticholinergic drug. On follow-up, she was doing well. Early diagnosis and long term follow-up is required in the patients having sacral agenesis with multidisciplinary team including urologist, as these patients may develop lower urinary tract symptoms, VUR and kidney injury at any time in their growth period.

Keywords: Bilateral pelvicalyceal system, Colostomy closure, Neurogenic bladder

CASE REPORT

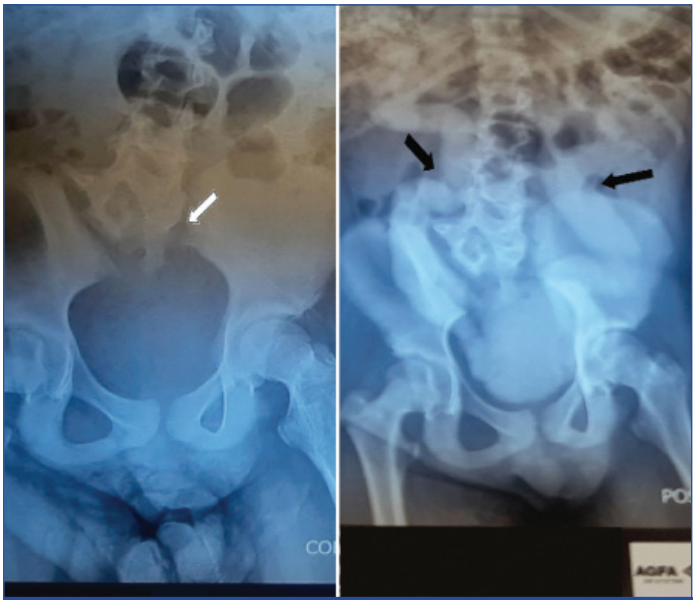
An 11-year-old girl presented to Urology Department with complaints of recurrent Urinary Tract Infection (UTI), persistent dribbling of urine and constipation for last one year. She was a known case of sacral agenesis with anal vestibule for which she was operated at the age of six months (anal transposition and transverse colostomy) and at the age of 14 months (colostomy closure). After which, she was lost to follow-up of primary treating physician. She was apparently well till the age of 10 years, after which she had recurrent episodes of UTI. There was no significant family history and no history of consanguineous marriage in the family. There was no maternal history of diabetes.

On examination, she was febrile, urinary bladder was palpable, buttocks were flattened [Table/Fig-1], external genitalia was excoriated and wet. Anal opening was in normal position with decreased anal tone and rectum loaded with hard stool (on per-rectal examination).



[Table/Fig-1]: Showing flattening of buttock.

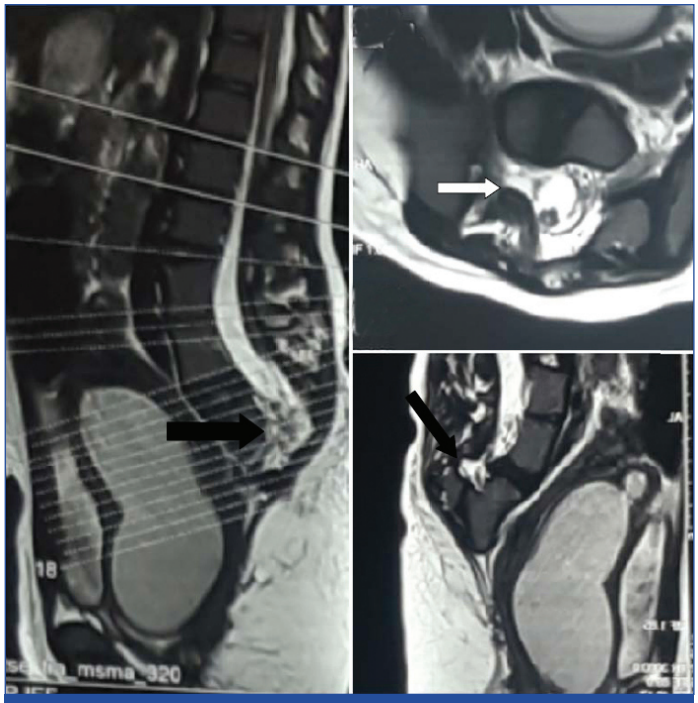
On evaluation, her blood analysis showed low haemoglobin (Hb-7.6 gm%), leukocytosis (14,500/cumm) and creatinine: 2.5 mg/dL. Ultrasonography (USG) of Kidney ureter and bladder (KUB) showed bilateral hydro-uretero-nephrosis and thickened irregular bladder wall with significant post void urine. Her urine analysis showed plenty of pus cells with *Escherichia coli* (colony count > one lakh per milliliter) on culture (antibiotic was given according to drug sensitivity). Sacral agenesis with absent ala of left side of sacrum seen in X-ray film and Voiding Cystourethrogram (VCUG) revealed bilateral grade 5 Vesico-Ureteric Reflux (VUR) as per International Reflux Grading System [Table/Fig-2]. Magnetic Resonance Imaging (MRI) of lumbosacral spine showed Type-III sacral agenesis with low lying cord with evidence of meningomyelocele [Table/Fig-3].



[Table/Fig-2]: Skiagram showing sacral agenesis (white arrow) and bilateral grade 5 VUR (black arrows).

On further evaluation with Urodynamic Study (UDS), she had detrusor overactivity and increased bladder capacity during the filling phase and strong detrusor contraction and obstructive pattern during voiding phase of micturition. After catheterization, her serum creatinine came down to normal level then she was advised for Clean Intermittent Catheterization (CIC) along with anti-cholinergic drugs (oxybutynin 5 mg/day) and a laxative.

The patient was followed-up three monthly on out-patient basis with serum creatinine, urine culture and USG KUB, which were



[Table/Fig-3]: T2 weighted images of spine showing sacral agenesis (White arrow) with evidence of meningomyelocele (Black arrow)

with in normal limits, apart from dilated bilateral pelvicalyceal system and ureters (due to VUR) on USG, which was also decreased when compared with previous USG images. Patient was doing well on CIC and anti-cholinergic drug, with no lower urinary tract symptoms.

DISCUSSION

Sacral Agenesis (SA) is defined as the absence of part or all of two or more vertebral bodies. It is a rare developmental anomaly of sacrum and frequently associated with bladder dysfunction [1]. Overall incidence of SA is 0.09-0.43% and occurs in about 0.1-0.25 per 10,000 pregnancy with its strong association with maternal diabetes mellitus [2]. It is mostly associated with other abnormalities, such as maldevelopment of the bones or joints of the lower limb and anorectal malformation [3]. SA was described as a part of spectrum of sacrococcygeal malformations, known as caudal regression syndrome [4]. SA may be associated with homeobox gene abnormalities and currarino triad [1,5,6].

Renshaw classification of SA:

Type I: Unilateral agenesis of sacrum or coccyx.

Type II: Iliac bone articulates with S1, distal sacrum not developed.

Type III: Total SA, iliac bones articulate with lowest lumbar vertebrae.

Type IV: Total SA, iliac bones fuse posteriorly [7].

Usually it has a bimodal presentation, with more than 75% presents in early infancy and the remainder detected between four and five years of age due to failed attempts of toilet training. Common urological presentations are voiding dysfunction and recurrent urinary tract infection [2]. The underlying lesion is usually overlooked, as they have normal sensation and minimal or no bony deformity in the lower limbs.

Due to total or partial absence of sacrum, injury occurs to the nerve fibres that pass through the bony canal of sacrum and supply the bladder , bowel, anus and sensory and motor fibres of lower limbs [8]. Patient may present with constipation, urinary incontinence, recurrent urinary tract infections and even renal injury as storage and voiding of urine occurs at abnormally high pressure due to neurogenic bladder [2]. Urodynamic study (UDS) may show either upper motor and/or lower motor type of bladder injury, but about 25% of patients may not find any sign of denervation [9]. VUR may be associated with both types of lesion but more due to detrusor overactivity. VUR can be diagnosed by VCUG. VUR is graded by International Reflux Grading System into five grades [10].

Here, authors present a known case of SA with vestibular anus, who lost follow-up of primary treating physician and ultimately presented to our Urology Department with recurrent urinary tract infection (UTI) due to bilateral grade 5 vesico ureteric reflux (VUR) with neurogenic bladder at the age of 11 year. She was managed with CIC and anti-cholinergic drugs. And she was doing well on follow-up.

In one retrospective study of 43 patients having SA, majority of the patients required CIC for the management of their neurologic bladder and none had developed end-stage renal disease [11]. In one case report by Sharma S et al simalar case was reported in a 7-year-old boy where radiograph of lumbosacral area of spine showed an abnormal sacrum [12]. Usually, treatment consist of anti-cholinergic for bladder overactivity or CIC with sympathomimetic for bladder underactivity. After initiation of treatment, urinary tract of these patients should be monitored on follow-up otherwise they may develop VUR or hydronephrosis leading to renal damage. These patients may require ureteric reimplantation (if high grade VUR present), augmentation cystoplasty with or without bladder neck continence surgery in order to maintain low bladder filling pressure and adequate bladder capacity to prevent renal damage in long run.

CONCLUSION(S)

Authors reported a case of a girl, who was initially diagnosed to have SA due anorectal malformation at the age of six months. Anorectal malformation corrected at the same time (six months of age). But after a prolonged period, she presented to the urology department with kidney injury due to recurrent UTI, on evaluation found to have neurogenic bladder with bilateral grade 5 VUR. The patient was fortunately not presented at this age with chronic kidney damage showing the importance of initial evaluation after diagnosis and regular follow-up of these group of patients by multidisciplinary team including paediatrician, neurologist and urologist.

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PLAGIARISM CHECKING METHODS: [Jain H et al.]
- Plagiarism X-checker: Feb 22, 2021
- Manual Googling: Apr 24, 2021
- iThenticate Software: Jun 10, 2021 (4%)

ETYMOLOGY: Author Origin

AUTHOR DECLARATION:
- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study? Yes
- For any images presented appropriate consent has been obtained from the subjects. Yes

Date of Submission: **Feb 16, 2021**
Date of Peer Review: **Apr 26, 2021**
Date of Acceptance: **May 21, 2021**
Date of Publishing: **Jul 01, 2021**