

# Sacral agenesis with isolated neurogenic bladder dysfunction

## Abstract

Sacral agenesis is a rare congenital vertebral anomaly which is often associated with bladder, bowel and lower limb neuromuscular dysfunction. An isolated bladder involvement is quite exceptional. Here we are reporting one such neglected case with isolated bladder involvement leading to chronic kidney disease.

**Keywords:** sacral agenesis, neurogenic bladder, caudal regression syndrome

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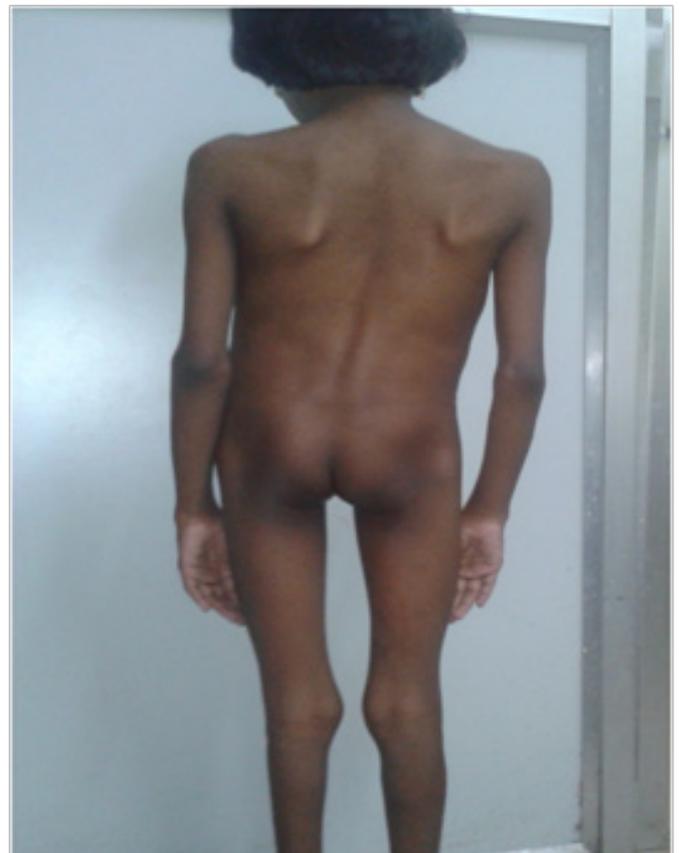
## Introduction

Sacral Agenesis (SA) is a rare congenital anomaly of the lower vertebral column, frequently associated with bladder dysfunction.<sup>1</sup> The cause of SA is still uncertain, but teratogenic factors may play a role as a clear association with insulin-dependent diabetes in the mother has been seen.<sup>2</sup> It may occur in isolation but usually it is associated with other abnormalities, such as anorectal malformation and mal development of the bones or joints of the lower limb.<sup>3</sup> Voiding dysfunction and recurrent urinary tract infection are common urological presentation.<sup>2</sup> However, kidney damage could occur if diagnosis is delayed and not intervened in time. Here we present a neglected case of sacral agenesis with neurogenic bladder leading to chronic kidney disease.

## Case report

An eleven year old girl was presented to with chronic constipation, persistent dribbling of urine and recurrent urinary tract infections since early childhood. She received treatment by local physicians but remained without proper response. On examination, child was a febrile and her bladder was palpable. Her buttocks were flattened (Figure 1) and external genitalia were wet and excoriated. Anal tone was found decreased on digital rectal examination and the rectum was loaded with hard stool. No definite neurological deficit was found. She was catheterised and stool evacuated with per rectal enema. Her blood analysis showed low haemoglobin (8.1gm %), leucocytosis and raised serum creatinine level (2.2mg %). Urine analysis showed plenty of pus cells and growth of E. coli. Ultrasonography suggested bilateral hydro ureteronephrosis (rt>>lt) with loss of cortico medullary differentiation on right kidney and thickened irregular bladder with significant post void residual urine. Tc-99m DTPA renogram shows poorly functioning right kidney and satisfactorily functioning left kidney. Complete sacral agenesis, 5th lumbar vertebral atrophy and scoliosis with convexity towards right side seen in the control film (Figure 2) while neurogenic bladder with bilateral grade V vesico-ureteric reflux revealed on Voiding phase cystourethrogram (Figure 3). Child was further evaluated with urodynamic study, which

suggested an overactive bladder in filling phase (Figure 4). Repeat blood analysis shows (Figure 4) normalisation of serum creatinine (1.2mg %). Her catheter has been removed and she has been placed on self clean intermittent catheterisation along with anti cholinergic (oxy butinine 5mg/daily) and laxative.



**Figure 1** Showing flattening of the buttock.



Figure 2 Complete Sacral agenesis, 5th lumbar vertebral atrophy.



Figure 2a Showing bilateral grade V vesico- ureteric reflux.



Figure 3 Complete Sacral agenesis, 5th lumbar vertebral atrophy.

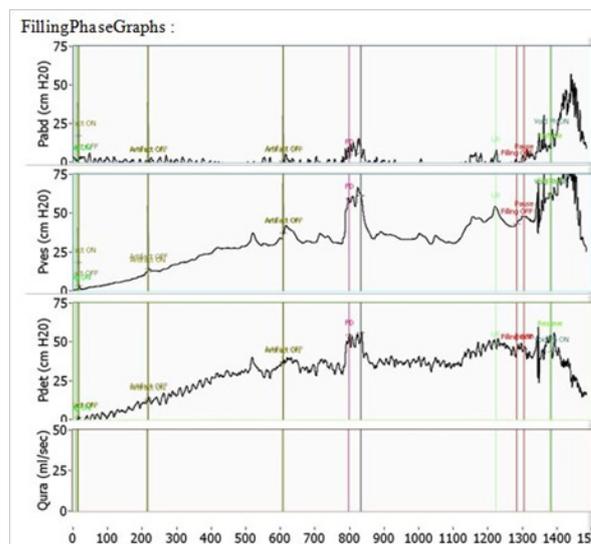


Figure 4 Cystometry showing overactive filling phase.

## Discussion

Sacral agenesis (SA) is a rare and severe sacral developmental abnormality.<sup>1</sup> It has been defined as the absence of part or all of two or more lower vertebral bodies. Insulin-dependent diabetes in the mother has been shown to be associated with sacral agenesis but in most of the cases cause is uncertain.<sup>2</sup> The presentation is usually bimodal, with more than three fourths presents in early infancy and the remainder discovered between 4 and 5 years of age.<sup>2</sup> Sacral agenesis may also be associated with abnormal development in other organs such as the anus/rectum, the bones and joints of the leg and spine.<sup>3</sup> When these all occur together, this is referred to as Caudal Regression syndrome.<sup>4</sup> Familial cases of sacral agenesis are seen to be associated with the Currarino syndrome (presacral mass, sacral agenesis, and anorectal malformation).<sup>5</sup>

Nerves from the spinal cord pass through a bony canal within the sacrum and exit to supply the bowel, anus, bladder, and to the muscles and sensory organs in the lower limbs. Due to absence of two or more bony segment of sacrum, this agenesis leads to injury to these nerve fibres.<sup>6</sup>

Renshaw classified SA in four types

- Type I (unilateral agenesis localised to sacrum or coccyx)
- Type II (partial agenesis with bilateral defects; the iliac bone articulates with S1, but the distal sacral elements fail to develop)
- Type III (total sacral agenesis; iliac bones articulate with the lowest lumbar element)
- Type IV (total sacral agenesis; iliac bones fuse posteriorly).<sup>7</sup>

However, the number of affected vertebrae does not seem to correlate with the type of motor neuron lesion present and the injury appears to be stable and rarely shows sign of progressive denervation.<sup>4</sup> Patient may present with constipation, faecal incontinence, urinary incontinence, recurrent urinary tract infections and even kidney damage as neurogenic bladder has to store and empty urine at abnormally high pressure.<sup>2</sup> Urodynamic evaluation usually reveals either upper motor or lower motor type of bladder injury, but about 25% of patient may not show any sign of denervation.<sup>4</sup> Vesico

ureteric reflux may be associated with both types of lesion but more with detrusor over activity.<sup>2</sup> Treatment usually consist of anti cholinergic for bladder over activity and CIC with sympathomimetic for underactive bladder.<sup>8</sup>

Present report is a neglected case of sacral agenesis who presented late at 11years of age. Constipation and urinary incontinence was main complaint and on evaluation she found to have type III sacral agenesis, overactive bladder with bilateral reflux disease leading to kidney injury. High index of suspicion and some clinical clues like flattened buttock and short gluteal folds,<sup>9</sup> are helpful for early diagnosis to avoid social stigma of urinary incontinence and to prevent kidney injury.

## Acknowledgements

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## Conflict of interest

The author declares no conflict of interest.

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