Case Report

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Sacral Agenesis with Neurogenic Bladder Dysfunction—A Case Report and Review of the Literature

Orthopaedics Section

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ABSTRACT

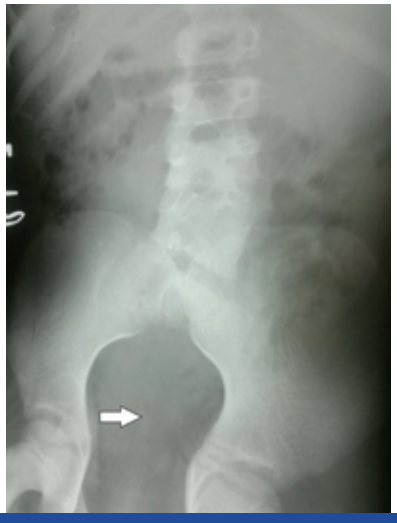
Sacral agenesis (part of the caudal regression syndrome) is a rare and severe sacral developmental abnormality. It is a congenital malformation of unknown aetiology with possible involvement of genetic and teratogenic factors. It is described by various degrees of developmental failure, the most extreme and rare being sirenomelia or mermaid syndrome. The associated malformations comprise anorectal, vertebral, urological, genital, and lower limb anomalies. Approximately 15-20% mothers of these children have insulin dependent diabetes mellitus. The case is being reported for its rarity and educative value because prognosis is good in isolated sacral agenesis.

Keywords: Anomalies, Caudal regression syndrome, Sirenomelia

CASE REPORT

A 7-year-old male child presented with poor urinary control since infancy with constant overflow incontinence. Since no organic cause could be found at previous examinations, the child was labelled as psychosomatic, a diagnosis that led to great unhappiness and extremely stressful domestic relationships. He was borne of a non consanguineous marriage, by normal vaginal delivery at term with breech presentation following an uneventful pregnancy of a non-diabetic mother. There was a history of congenital malformation in the form of bilateral congenital talipes equinovarus at birth which was corrected by serial applications of plaster cast upto the age of one and half year. There was no neuro developmental delay.

On examination, patient's weight and height were appropriate for age. Patient had a normal gait. There was continuous dribbling of urine with extensive excoriation of the area, and a dimple over sacral area. Nervous system examination was normal except for sluggish ankle jerk on both sides and anal sphincter hypotonia. Musculoskeletal examination was normal. Radiograph of lumbo-sacral area of spine showed an abnormal sacrum [Table/Fig-1] with type 4 anomaly as per Renshaw classification [Table/Fig-2]. Normal ultrasonography ruled out any associated visceral anomalies except for residual urine (approx.250 cc) in bladder. This along with anal hypotonia confirmed the presence of a mixed upper and lower motor neurogenic bladder with normal sensation. Lab investigations including complete blood count, renal function tests and serum electrolytes were normal. Urine culture was negative even after 72 hours of incubation. Child was sent to urologist for urinary diversion and uro-dynamic study.



[Table/Fig-1]: Radiograph showing sacral agenesis (Arrow)

Type I	Partial or total unilateral sacral agenesis
Type II	Partial sacral agenesis with bilaterally symmetrical defect a normal or hypoplastic sacral vertebra, and a stable articulation between the ilia and first sacral vertebra
Type III	Variable lumbar and total sacral agenesis, with the ilia articulating with the sides of lowest vertebra present
Type IV	Variable lumbar and total sacral agenesis, with the caudal endplate of the lowest vertebra resting above either fused ilia or an iliac amphiarthrosis

[Table/Fig-2]: Renshaw classification of sacral agenesis

DISCUSSION

Sacral agenesis (sacral hypoplasia) can occur in isolation or as a part of caudal regression syndrome and involves abnormal fetal development of vertebral column and spinal cord [1]. It is a rare anomaly with a very low incidence (approximately 1 per 25,000 live births). By 4th week of gestation formation of the sacrum/lower back and corresponding nervous system is usually nearing completion. According to some studies perturbations of mesoderm specification, epithelial-mesenchymal transition, and mesodermal cell migration can lead to such structural birth defects [2].

In 1961, Duhamel coined the term "syndrome of caudal regression" for this and subsequently Smith and Karrer suggested that caudal vertebrae and spinal cord anomalies occur in 40 to 50% of children with anorectal malformations. The aetiology of CRS is still unclear but it has a well-known association with insulin-dependent maternal diabetes mellitus [3]. The only aetiologic agent associated with the CRS were reported in mothers exposed to organic fat solvents during the first months of their pregnancies [4]. Apart from these teratogenic agents such as exposure to retinoids, insulin, embryonal trauma, severe fluctuations in temperature, vitamin deficiencies, lithium salts, radiation, stress, alcohol, amphetamines, and trypan blue have all been implicated as causes of caudal agenesis [5]. Familial cases suggest genetic cause with a recurrence risk of 5% in families that already had an affected child. CRS would be transmitted as either dominant or recessive characteristics. The dominant inherited sacral agenesis known as Currarino syndrome is correlated with the disease causing HLXB9 gene, located at 7q36 [6].

In our case, there was no history of maternal diabetes or teratogenic substance intake during gestation with negative familial history. Caudal regression syndrome may present as absent coccyx without

neurological sequelae, to sacral or lumbosacral agenesis [7] along with other system involvement [Table/Fig-3].

Renshaw has classified sacral agenesis in four types [Table/Fig-2] [8]. Usually there is no correlation between level or degree of the bony abnormality and associated neurological deficit, the motor deficit being always more pronounced than the sensory deficit so that inspite of intact perianal sensation there may be bladder or anal sphincter involvement [9].

Orhopaedic	Congenital hip or knee dislocation
	Coxa vara
	Talipes equinovarus and calcaneovalgus
	Absent or atrophic extremities
	Myelomeningocele
	Absent, dysplastic or fused vertebra
	All variants of anorectal malformations
Gastro intestinal	Inguinal and umbilical hernias
	Duodenal atresia
	Malrotations and situs inversus
	Vesico-ureteral reflux
Urological	Congenital hydronephrosis
	Hypospadias
	Cryptorchidism
	Duplication of collecting system
	Renal agenesis
Miscellaneous	Tracheo-oesophageal fistula
	Hydrocephalus
	Strabismus
	Congenital heart disease

[Table/Fig-3]: Congenital anomalies associated with partial or complete sacral agenesis

Sometimes patient may present with dribbling of urine or chronic urinary infection secondary to vesico-ureteral reflux without any evident signs suggesting sacral agenesis. Look out for flattened gluteal cleft, gluteal dimpling, a sacral lipoma or myelo-meningocele with any of the associated orthopaedic, anorectal or other congenital defects [Table/Fig-3] suggests agenesis which should be confirmed by an antero-posterior radiograph of the spine [9].

Antenatal ultrasound is important for diagnosis of fetal CRS [10]. Sacral or lumbosacral agenesis in combination with marked hypoplasia of lower extremities is diagnostic of CRS. The measurements of fetal crown-rump length of CRS foetuses in the first trimester have been reported to be commonly shorter per week than normal foetuses [11]. Before 19 weeks of pregnancy, the diagnosis of sacral agenesis by ultrasound is difficult because the sacrum is not calcified enough.

Depending upon underlying pathology bowel or bladder deficiencies is quite common in sacral agenesis. Imperforate anus may require a colostomy while incontinence may be managed by cathetrisation. Because of the very high incidence of vesicoureteral reflux all patients with sacral agenesis should also have a voiding cysto-urethrogram taken routinely. Intermittent clean self catheterisation along with uropharmacological manipulation is preferred mode of treatment for neurogenic bladder [11]. Occasionally if deformities of the knees, legs or feet would prove unresponsive to corrective action, a disarticulation is done at the knee for those who have bent knee positions and webbing between thigh and calf to enable more ease of mobility and better seating. In some children with sacral agenesis and non-functional lower limb deformities, amputation and subsequent wheelchair or prosthetic mobilisation is the preferred way of management. Some children are known to adapt very well to this condition by mobilising about with their hands. No cognitive impairment is associated with this disability. Adults with this disability are known to live independently, attend college, and have careers in various fields. Spencer West from Canada, born with sacral agenesis and subsequent bilateral above knee amputee was able to reach summit of Mt Kilimanjaro just with his hands [11].

CONCLUSION

The diagnosis of sacral agenesis is often overlooked as happened in our case. As the sacral agenesis is associated with significant morbidity, a delay in diagnosis can lead to progressively deteriorating urinary function and chronic urinary tract infections. Hence, we suggest that presence of any associated anomalies and urinary dysfunction should attract our attention to underlying possibility of sacral agenesis.

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