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ORIGINAL ARTICLE

Sacral agenesis and fecal incontinence: how to increase the index of suspicion

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Abstract

Purpose Sacral abnormalities range from missing the coccyx, a few sacral vertebrae, or hemi-sacrum, to complete absence with fused iliac bones. The purpose of this study was to review the association between sacral agenesis and fecal incontinence to help inform patient prognosis.

Methods A retrospective review was performed of patients who presented for bowel management due to sacral agenesis at a tertiary care children's hospital between 2016 and 2017 ($n = 10$). Data collection included: gender, time of diagnosis, sacral ratio, and associated anomalies. Patients with anorectal malformation and sacrococcygeal teratomas were excluded.

Results Four patients were female. Seven patients had a delayed diagnosis ranging from 22 months of age to 9 years. Most common symptoms included failure of age-appropriate toilet training and severe diaper rash. The sacral ratio was zero (6), 0.3 and 0.4 (2), and hemi-sacrum (2). Associated anomalies were present in five patients.

Conclusion Sacral abnormalities should be suspected in patients who present with early severe diaper rash and those who fail to toilet train. An abdominal radiograph can evaluate the sacrum, when the sacral ratio is 0.4 or less, parents should be counseled regarding fecal incontinence and neurogenic bladder.

Keywords Sacral agenesis · Fecal incontinence · Sacral abnormalities · Caudal regression

Introduction

Sacral agenesis is a rare congenital disorder with a wide spectrum in terms of severity. Incidence has been reported as 0.01–0.05 per 1000 live births. There are four types of sacral agenesis. Type I includes total or partial unilateral sacral agenesis. Type II is characterized by partial, bilateral symmetric defects that are in stable articulation between ilia and normal or hypoplastic S1 vertebra. Type III includes variable lumbar and total sacral agenesis with the ilia articulating with the lowest vertebra. Type IV is represented by variable lumbar and total sacral agenesis where the caudal endplate of the lowest vertebra rests above the fused ilia [1]. Sacral agenesis or caudal regression syndrome can also be associated with other abnormalities involving the nervous system, genitourinary system, gastrointestinal system and musculoskeletal system.

There is a paucity of literature with varying reports regarding the association between sacral agenesis and fecal incontinence [2]. When reviewing the associated symptomatology regarding lower extremity weakness, urinary symptoms, and fecal incontinence, one study found that 34% of the patients with sacral agenesis had fecal incontinence and that it was more prevalent in patients with imperforate anus and a lower sacral ratio [3]. However, another study reported a higher prevalence with 84% of patients with sacral agenesis reporting fecal incontinence [4]. Furthermore, in a study performing rectal manometry on patients with sacral agenesis, all of the patients were incontinent of stool. Additionally, they found that patients with total sacral agenesis had lower anal squeeze pressure and blunted sensation of rectal distention [5]. With such variability in the literature regarding the prevalence of fecal incontinence in relation

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to sacral agenesis, it is difficult to provide families with prognostic information if the diagnosis of sacral agenesis is made. The purpose of this study was to review the association between sacral agenesis and fecal incontinence as well as other anomalies.

Methods

Study design

A retrospective chart review was performed after obtaining IRB approval (17-1636). Inclusion criteria included patients with the diagnosis of sacral agenesis at a tertiary care children's hospital between 2016 and 2017 ($n = 10$). Those with anorectal malformations and sacrococcygeal teratomas were excluded.

Variables

Variables collected included gender, race, age, diagnosis, age at diagnosis, sacral ratio, presence of fecal or urinary incontinence, presence of neurological deficits, radiologic workup, procedures, complications, and bowel management regimen.

Statistical analysis

Descriptive statistics included proportions for categorical variables and median with interquartile range for continuous variables. Due to the small sample size, further statistical analysis was not performed.

Results

Patient characteristics

Most patients were male (6/10). The median age at diagnosis was 4.5 (range 0–9) years with the majority of patients (7/10) experiencing delayed diagnosis with the age of delayed diagnosis ranging from 22 months to 9 years of age. Reviewing the sacral ratio, 6/10 patients had a sacral ratio of zero, 2/10 patients had a hemi-sacrum, and 2/10 patients had a measurable ratio of 0.3 and 0.4 (Fig. 1). The most common symptoms included failure of age appropriate toilet training (4/10), severe diaper rash (3/10), and constipation in a patient that was still in diapers (1/10). The remaining 2/10 patients were appropriately diagnosed and referred for bowel management prior to development of symptoms. MRI was performed in 8/10 patients. The most common findings on MRI were blunt conus (6/8) and tethered cord (2/8).

Associated anomalies

All patients experienced fecal incontinence and neurogenic bladder. Other anomalies were present in 5/10 patients. Neurological anomalies included agenesis of corpus callosum (1/10), developmental delay (1/10), tethered cord (2/10), and meningocele (1/10). Urological anomalies included undescended testicle (1/10), vesicoureteral reflux (VUR) (3/10), and a single kidney (1.10). Cardiac anomalies included transposition of great arteries (TGA) (1/10) and VSD (1/10). Musculoskeletal anomalies included bilateral club foot (4/10), bilateral hip dysplasia (1/10) and microtia (1/10) (Table 1).

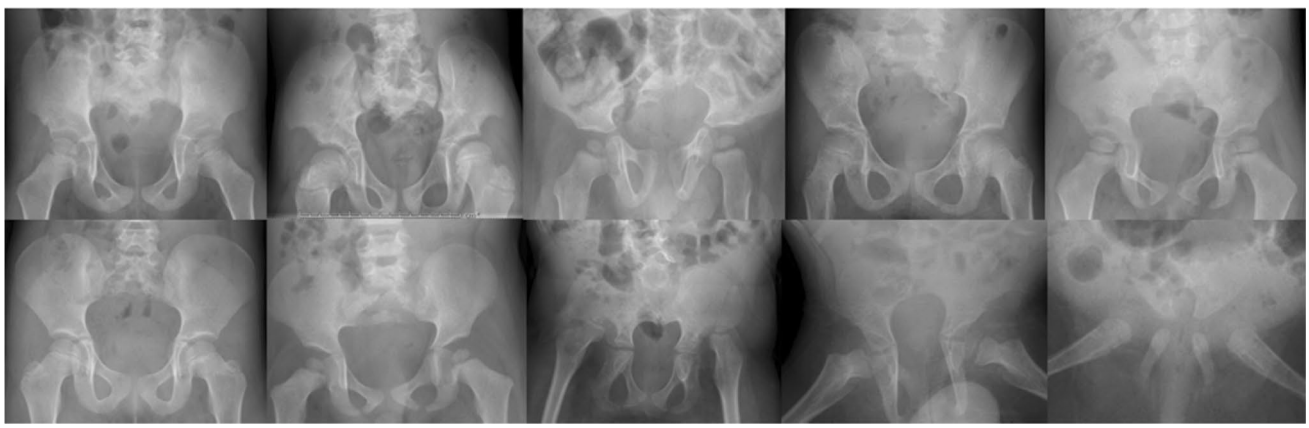

Fig. 1 Abdominal radiographs of patients with sacral agenesis

Table 1 Associated anomalies in patients with sacral agenesis

Anomaly	Proportion of patients
Fecal incontinence	10/l0 (100%)
Neurological	
Blunt conus	6/10 (60%)
Tethered cord	2/10 (20%)
Corpus callosum agenesis	1/10 (10%)
Developmental delay	1/10 (10%)
Meningocele	1/10 (10%)
Urological	
Neurogenic bladder	10/10 (100%)
Vesicoureteral reflux	3/10 (30%)
Single kidney	1/10 (10%)
Undescended testicle	1/10 (10%)
Musculoskeletal	
Club foot	4/10 (40%)
Hip dysplasia	1/10 (10%)
Microtia	1/10 (10%)
Atrophic legs	3/10 (30%)
Cardiac	
TGA	1/10 (10%)
VSD	1/10 (10%)

Treatment

Reviewing management of fecal incontinence, eight patients are artificially clean for stool with daily rectal enemas (three of them have already received a Malone for antegrade enemas), and 2/10 patients are waiting to start bowel management. In terms of urinary incontinence, 3/10 patients were managed with a Mitrofanoff, 2/10 patients perform clean intermittent catheterization through the urethra, and 1/10 patients had a vesicostomy.

Discussion

This study highlights that all patients with sacral agenesis have fecal incontinence. Furthermore, most of these patients experienced delayed diagnosis of fecal incontinence, as the association between fecal incontinence and sacral agenesis was not recognized. The most common symptoms that eventually led to diagnosis were severe diaper rash and failure to toilet train by the appropriate age. In addition to fecal incontinence, all these patients experience urinary incontinence. The most common other anomalies include clubfoot, vesicoureteral reflux, and tethered cord. Management of incontinence included intermittent catheterization per urethra or Mitrofanoff for urinary symptoms or enemas per rectum or Malone for bowel symptoms.

Previous studies regarding the relationship between fecal incontinence and sacral agenesis are limited. Other studies have similarly demonstrated the problem of delayed diagnosis in this patient population. These studies also note that a significant proportion of their population present with associated musculoskeletal abnormalities and VUR. However, these studies did not examine the prevalence of fecal incontinence [6, 7]. The importance of the sacral ratio in determining fecal as well as urinary continence prognosis has been previously described [8, 9]. This is further supported in the current analysis where the highest sacral ratio was 0.4 with most patients having a sacral ratio of 0.

By recognizing the association between sacral agenesis and fecal incontinence, these patients may be diagnosed earlier and appropriately referred to a bowel management program prior to the development of significant gastrointestinal symptoms. Furthermore, the diagnosis of sacral agenesis may allow for appropriate prognostic counseling with the family regarding fecal and urinary continence.

Limitations of this study include its retrospective nature and small sample size. However, with the limited available literature regarding this subject, this study provides valuable insights into the anomalies associated with sacral agenesis and the importance of recognizing the relationship between sacral agenesis and these anomalies for appropriate patient management. Furthermore, potential confounders of fecal incontinence were addressed by excluding patients with anorectal malformations and sacrococcygeal teratomas.

Conclusion

Patients presenting with severe diaper rash or failure to toilet train at the appropriate age should be suspected of having a sacral abnormality. This can easily be assessed through sacral radiographs. A sacral ratio of 0.4 or less correlates with fecal incontinence and neurogenic bladder; patients should be referred to a bowel management program and urological evaluation. Patients with complete sacral agenesis (fused iliac bones) are non-ambulatory.

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