CASE SEREISE ARTICLE

SPINAL AND VERTEBRAL ANOMALIES ASSOCIATED WITH ANORECTAL MALFORMATIONS

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Abstract

Objective

The associations between imperforate anus and spinal and vertebral abnormalities and neurologic deficits are well recognized; these neurologic deficits have been considered static rather than progressive. However, recent experience indicates that some patients may develop progressive neurologic problems due to spinal cord lesions that are amenable to neurosurgical correction.

Materials & Methods

The medical records of 105 patients with imperforate anus, operated on by us, were retrospectively reviewed from 1996 to 2005. Patient's sex, anorectal type lesion and vertebral or spinal anomalies were determined by ultrasound, excretory urography, voiding cystouretherography (VCUG) and lumbosacral x-ray.

Results

A hundred and five cases, consisting 48 (45.7%) boys and 57 (54.3%) girls, with anorectal malformations were studied; 70 patients were in high and intermediate type level, and totally 25 patients (35.7%) with spinal and vertebral anomalies were found in this group.

Conclusion

All patients with anorectal malformations (ARM) should be investigated for spinal and vertebral anomalies to improve treatment outcomes in ARM.

Keywords: Anorectal malformations, spinal, vertebral anomalies

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Introduction

Anorectal malformations (ARM) are a complex group of malformations diagnosed at the time of birth, because of either the absence or an ectopic location of anus (1). The incidence is approximately 1:5000 live births and they are seen more often in boys than in girls(2). The broad spectrum of ARM includes anal atresia, anal stenosis, ectopic anus, congenital anal fistula, and persistence/exstrophy of the cloaca (3).

Anal atresia is the most frequent of congenital anal anomalies; it is a complex condition that can be subdivided into high, intermediate, and low atresia according to the level of termination of the rectum or anal canal in relation to the levator ani muscle(4). Anorectal malformations are associated with several other congenital anomalies, involving most commonly the genitourinary system and vertebral column and also include segmentation anomalies and sacral agenesis (5). The vertebral anomalies have been associated with occult dystrophic lesions of the

spinal cord (occult myelodysplasia), which may result in cord tethering (6,7).

Material & Methods

All the children diagnosed with congenital anorectal malformations in Mofid children's hospital from 1996 to 2005 were studied retrospectively. Case records and imaging studies of 105 children were reviewed. Patient's gender, anorectal lesion level and the presence of vertebral or spinal anomalies were determined; the level of anorectal lesion was determined by radiography evaluation.

In 70 (66.7%) patients who had intermediate or high level anorectal lesions, ultrasound, excretory urography, VCUG and lumbosacral x-rays were performed. Those patients suspected of having associated spinal anomalies were evaluated with ultrasound, vertebral x-ray and MRI. Statistical analysis was performed using SPSS v 13 software (Apache Software Foundation, Chicago, Illinois).

Results

One hundred and five patients 48 (45.7%) boys and 57 (54.3%) girls) with ARM were enrolled in this study; 70 (66.7%) of these had high and intermediate types of this anomaly, and 35 (33.3%) low type (figure 1). All 25 (35.7%) of spinal and vertebral anomalies in our patients were in high and intermediate type of imperforate anus (70 cases) (Table1). Table 2 demonstrates the different spinal and vertebral anomalies in the 25 (35.7%) patients (figure 2).

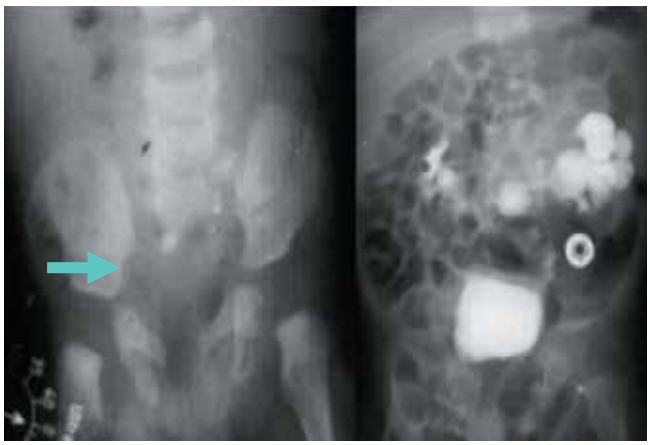

Figure 1: Excretory urogram showing partial agensis of sacrom in one of our patients with high imperforate anus.

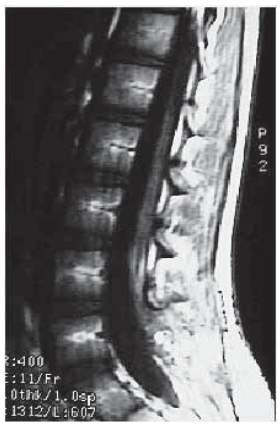

Figure 2: MRI of tethered cord in one of the studied patients

Table 1: ARM level according to gender in the 105 patients.

Gender (%)	Low		High+ intermediate	
Male	10	40	38	54.2
Female	25	60	32	45.8
total	35	100	70	100

Table 2: Spinal/Vertebral anomalies in70 patients with high/ intermediate type of ARM

Anomaly	Number	Percentage
Sacral Agenesis	8	32
Spina Bifidia	4	16
Hemi Sacrum	2	8
Myelomeningocele	1	4
Tethered Cord	5	20
Neurovesical Dysfunction(NVD)	5	20
total	25	100

Discussion

Imperforate anus is a complex maldevelopment of the anorectal region. This anomaly represents failure of the terminal hindgut to develop and the incomplete division of the cloaca by the urorectal septum that separates the ventral urogenital sinus from the dorsal anorectal canal (1).

Additional congenital anomalies are often present in patients with ARM, coexisting anomalies that account for the high morbidity and mortality associated with this condition (8). There is wide variation in the type of ARM and the range of associated anomalies (9). The incidence of associated vertebral and spinal anomalies reported in ARM subjects, varies from 16.67% to 38.3% in different series (10). The incidence of vertebral and spinal anomalies in our study was 35.7% which is similar to the data reported above.

Varying degrees of sacral abnormalities are commonly present in patients with imperforate anus, ranging from mild changes of sacral dysplasia (sacral stubbiness, sacral scoliosis) to varying degrees of sacral segmental agenesis (10).

The importance of the sacral defect is not in its need for spine support but in it's coexistence with defective bladder innervations, and poor development of the levator Ani sling (11, 12). In 3 of our patients there was neurovesical dysfunction, too.

Williams and Nixon (13) have stressed the importance of sacral dysplasia in the diagnosis of neurogenic bladder with or without vesico-ureteral reflux, commenting on the high incidence of imperforate anus in this group of patients (14).

Despite the lateral sacral study shows the abnormality, actual total absence of the sacrum was not present in any of our patients. The chance of an abnormal urinary tract is markedly raised in patients with imperforate anus who have sacral dysplasia (5). Specifically excluded from this definition of sacral dysplasia is nonosseous fusion of the upper sacral segments (15). Of the 25 patients in our study, 8 (32 %) had sacral dysplasia (spina bifida occulta).

Lumber anomalies are less emphasized in discussions of spine defects in patients with imperforate anus (4). Coexistent urologic defects are too mild as compared to those with sacral defects alone (3,12,16). In study of 104 ARM subjects, Mittal et al, found associated spinal anomalies in 10% and vertebral anomalies in 34.3 % of the subjects(16). In our study there were 8 patients with sacral agenesis.

Published prevalence of associated anomalies varies considerably because of the following: Differences in case definition and inclusion/exclusion criteria; length of time after birth that cases are examined; variability of clinical expression of associated anomalies; knowledge and technology available to produce syndrome delineation; selection of patients, sources of ascertainment, and sample size; and true population differences and changes in frequency over time (7).

In conclusion, because of the high incidence of spinal cord lesions in patients with coexisting anorectal and sacral anomalies, routine screening for spinal cord is recommended to improve treatment (17).

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