Anorectal Malformations and Down's Syndrome

By R. Torres, M.A. Levitt, J.M. Tovilla, G. Rodriguez, and A. Peña
New Hyde Park, New York, Bronx, New York

Background/Purpose: Down's syndrome is a common association in patients with anorectal malformations. The purpose of this study was to determine whether the anorectal defect in patients with Down's syndrome had specific characteristics and whether the presence of Down's syndrome represented a serious detriment to the patient's functional prognosis.

Methods: Nine hundred eighty-seven patients with anorectal malformations were studied retrospectively. Twenty patients (2%) had Down's syndrome. Nineteen of these (95%) had the same specific type of anorectal defect: imperforate anus with no fistula. This defect has a good prognosis, the rectum is located about 2 cm above the perineal skin, the sacrum is normal, and the sphincter mechanism is good. For comparison, a group of 34 patients with the same defect but without Down's syndrome was also studied. All patients were operated on via posterior sagittal approach by the same surgeon.

Results: Imperforate anus without fistula occurs in 5% of all patients with anorectal malformations and in 95% of those patients who also suffer from Down's syndrome. The characteristics of the defect were the same in both groups of patients, and surprisingly, the prognosis was good in both groups (80% to 96% of patients had voluntary bowel movement, 100% had urinary continence).

Conclusions: The association of Down's syndrome with imperforate anus without fistula is not coincidental. This particular benign defect can be predicted to occur in most patients with Down's syndrome. The presence of Down's syndrome in cases of anorectal malformations should not be a contraindication to repairing the imperforate anus and to closing the colostomy.

J Pediatr Surg 33:194–197.

INDEX WORDS: Down's syndrome, anorectal malformation, imperforate anus, fistula.

ANORECTAL MALFORMATIONS seem to be more frequent in patients with Down's syndrome,[1-10] with an incidence from 0.36% to 2.7%.[3,5,6,8,10] In series of patients with anorectal malformations, Down's syndrome is a commonly associated condition, much more frequent than the 0.15% incidence of Down's syndrome found in the general population[1,4,7-9] (Table 1).

We reviewed our series of 987 patients with anorectal malformations to learn more about this association and to answer several questions: (1) What is the incidence of Down's syndrome in this patient population? (2) Is there a predominant type of anorectal defect in Down's patients? (3) Do patients with Down's syndrome and imperforate anus have certain clinical features and associated anomalies that differ from other patients with anorectal malformations? (4) Do children with Down's syndrome differ in their functional prognosis for fecal and urinary continence?

MATERIALS AND METHODS

Of 987 patients with anorectal malformations in the senior author's personal series, 20 patients with Down's syndrome were identified and their cases analyzed for specific type of defect, quality of sacrum, perineal musculature, associated anomalies, and bowel and urinary function. All patients were operated on via posterior sagittal approach. For comparison, another group of patients ($n = 34$) without Down's syndrome and with the same specific type of defect was analyzed using the same parameters. The previously published classification of anorectal malformations was used.[11]

To evaluate the sacrum, the anterior-posterior and lateral-sacral ratios were used. These are derived by measuring the radiological image of the sacrum and comparing its size with fixed bony structures of the same patient's pelvis.[11] Children with anorectal malformations show a spectrum of values; lower ratios represent different degrees of sacral hypodevelopment and are associated with defects that have poor functional prognosis.

The perineal muscles were assessed by direct visualization during the operation, with and without electrical stimulation.

Bowel control was assessed in three areas: the presence of voluntary bowel movements, fecal soiling, and constipation. Voluntary bowel movements were defined as the act of feeling the urge to use the toilet to have a bowel movement, the capacity to verbalize it, and the ability to hold the bowel movement until the bathroom is reached. Soiling was defined as the involuntary leaking of small amounts of stool that leads to smearing of the underwear. Constipation was considered the incapacity to empty the rectum spontaneously (without help) on a daily basis.

Patients with urinary incontinence were those patients who were unable to hold their urine. Patients with nocturnal enuresis were

From the Division of Pediatric Surgery, Department of Surgery Long Island Jewish Medical Center, New Hyde Park, NY. The Clinical Campus of the Albert Einstein College of Medicine, Bronx, NY.

Presented at the 28th Annual Meeting of the American Pediatric Surgical Association, Naples, Florida; May 18-21, 1997.

Address reprint requests to Alberto Peña, MD, Chief, Division of Pediatric Surgery, Schneider Childrens Hospital, Long Island Jewish Medical Center, The Long Island Campus for the Albert Einstein College of Medicine, New Hyde Park, NY 11040.

0022-3468/98/3302-0008$03.00/0

Table 1. Incidence of Down's Syndrome in Patients With Anorectal Malformations

Study	No. of Patients With Anorectal Malformations	No. of Patients With Down's Syndrome	Incidence (%)
Buchin et al[4]	187	5	2.6
Joseph et al[7]	88	7	8
Gross[9]	198	8	4
Present study	987	20	2

considered in the continent group.[11] Only patients older than 3 years of age underwent investigation for functional status.

RESULTS

There were 20 patients with Down's syndrome in this series, a 2% incidence, with seven girls and 13 boys. Patients ranged in age from 2 months to 10 years, with a median age of 23 months. Nineteen of 20 patients (95%) had imperforate anus without fistula. One patient had a rectourethral bulbar fistula. There were 34 patients without Down's syndrome but with the same anorectal defect of imperforate anus without fistula (control group). Thus, the incidence of patients without fistula (with and without Down's) in our overall patient population was 5.3%. Of those patients without fistula, 36% (19 of 53) had Down's syndrome. The rectal pouch, in all cases, was found to be located within 2 cm of the perineal skin. Figure 1 is a diagram of imperforate anus without fistula.

Sacral measurements were available in 10 cases; the average lateral-sacral ratio was 0.66, and the average anterior-posterior ratio was 0.59. In the control group (imperforate anus without fistula and no Down's syndrome) 26 patients had available lateral-sacral films with an average ratio of 0.68. Seventeen of these patients had anterior-posterior sacral films, with an average ratio of 0.68. There was no statistically significant difference between the two groups. These ratios were similar to the ones obtained for patients with rectourethral bulbar fistula.[11]

Nineteen of 20 (95%) patients had well-developed perineal muscles. One patient had a flat bottom. In the control group, 28 of 30 patients (93%) had good perineal muscles. The difference was not statistically significant between the two groups.

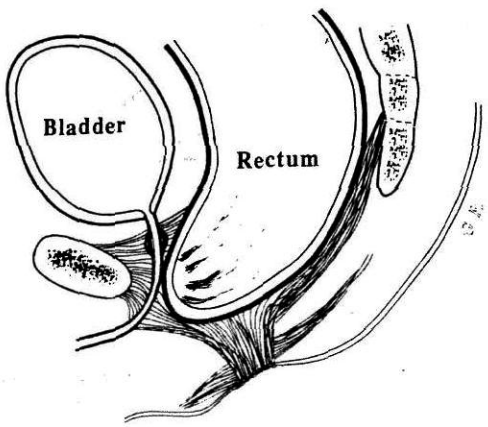

Fig 1. Imperforate anus without fistula.

Table 2 shows the associated anomalies present in the group of patients with Down's syndrome and in the control group. In the Down's group, cardiac anomalies were present in 50%. There were four patients with atrioventricular canal, two with ventricular septal defect (VSD), two with Tetralogy of Fallot, one with atrial septal defect, and one with patent ductus arteriosus (PDA). Urologic anomalies were present in 25% of patients (two with undescended testis, two with hydronephrosis, and one with absent kidney).

In the control group, 9% had cardiovascular anomalies (two with PDA, one with VSD) and 29% had a urologic anomalies (two with undescended testis, two with hydronephrosis, one with an absent kidney, two with hypospadias, two with vesicoureteral reflux, and one with ureteral stenosis). The two groups differed significantly in the presence of cardiac anomalies but were not different statistically in the presence of urologic anomalies.

Interestingly, in the control group, we noted associated syndromes in five patients, including Apert syndrome, Opitz syndrome, absent corpus collosum, mental retardation, and cleft lip and palate.

In the group of patients with Down's syndrome, clinical information concerning continence was available for 15 patients. One patient was lost to follow-up, and four patients were less than 3 years old. Voluntary bowel movements were present in 80% of patients, occasional soiling in 46%, constipation in 46%, and urinary continence in 100% (Table 3).

Clinical data regarding continence was available for 25 of the 34 patients in the control group; 96% had voluntary bowel movements, 48% had occasional soiling, 48% had constipation, and all patients had urinary continence (Table 3). There was no statistically significant difference between these two groups.

DISCUSSION

Table 1 shows the incidence of Down's syndrome in patients with anorectal malformations. The present study's 2% incidence is different from these, which range from 2.6% to 8%, and may be more representative of the actual incidence because of the sample size.

Table 2. Associated Anomalies Found in Patients With Imperforate Anus Without Fistula

	Patients With Down's Syndrome	Patients Without Down's Syndrome	*P* Value
Cardiovascular	10/20 (50%)	3/34 (9%)	<.001
Genitourinary	5/20 (25%)	10/34 (29%)	NS

Abbreviation: NS, no statistically significant difference.

Table 3. Bowel and Urinary Continence in Patients With Imperforate Anus Without Fistula

	Patients With Down's Syndrome	Patients Without Down's Syndrome
Voluntary bowel movement	12/15 (80%)	24/25 (96%)*
Soiling	7/15 (46%)	12/25 (48%)*
Constipation	7/15 (46%)	12/25 (48%)*
Urinary continence	15/15 (100%)	25/25 (100%)*

*No statistically significant difference.

Imperforate anus without fistula is a very specific and unique defect with good functional prognosis. The rectum is located more or less in the same place, the sacrum is basically normal, and the sphincters are very good. In addition, the association with Down's syndrome is not coincidental.

From an English-language literature review of reports from 1966 to 1996, one paper specifically concerned imperforate anus and Down's syndrome.[1] It is difficult to make comparisons between that study and this series because the investigators used a different classification, describing a "low-lying rectal pouch without a genitourinary or perineal fistula" in all of their patients. The difference between those investigators' findings[1] and ours may be simply semantic variation. When they talk about a low-lying rectal pouch it could be interpreted as a "low" imperforate anus according to the older classification. The therapeutic implication of that name usually was that the patient could be operated on primarily via a perineal approach, without a protective colostomy. Such an approach could place the patient at risk because they could have a bulbar urethral fistula that radiologically may look like a low lesion. We have seen patients with this kind of fistula approached perineally by other surgeons, and they left the fistula untouched. In fact, most patients with imperforate anus without fistula have their rectal pouch located at the same level as those with rectourethral bulbar fistula. In addition, the rectum is intimately attached to the posterior urethra,[12] and thus the repair of this defect is as technically demanding as in those cases with a fistula. It is therefore safer for the patient to have a protective colostomy.

In the study by Black et al,[1] the level of the rectal pouch was determined by physical examination, invertogram, or ultrasound scan in the majority of their patients. Distal colostography was performed in only three of the patients. In our series, the anorectal diagnosis and the height of the defect in all patients were determined through use of high-pressure distal colostography.[13] Augmented-pressure colostography is the most reliable method of determining the true height of the defect, because the rectum passes through a funnellike muscular structure, which is usually contracted, resulting in compression of the rectal lumen. If not enough hydrostatic pressure is applied during the injection of contrast material through the mucous fistula, inadequate distension of the distalmost part of the rectum will result, and a false impression of a higher defect will occur. Other methods of determining the rectal pouch height are unreliable. The importance of this procedure cannot be overestimated because improperly performed distal colostography may fail to define the distalmost rectal segment and any associated rectourinary fistula. Figure 2 shows an example of a distal colostogram, with adequate distension of the rectal pouch, demonstrating a defect with no fistula.

The anorectal defect that occurred in our patients with Down's was almost uniformly imperforate anus without fistula, which is in agreement with Black and Sherman's series.[1] Clinicians must suspect that patients with Down's syndrome will most likely have this type of anorectal defect with a rectal pouch about 2 cm from the perineal skin, which can be repaired via the posterior sagittal approach without laparotomy.

The pattern of associated anomalies suggests that the cardiovascular malformations were related to the presence of Down's syndrome, a known association,[14-16] and not to the anorectal malformation. The urologic anomalies seem, however, to be related to the presence of imperforate anus, a known association,[17] because the incidence was similar in the Down's syndrome patients and in the control group.

The association of imperforate anus without fistula and rare syndromes, which occurred in five patients without Down's syndrome, is an interesting one. The absence of fistula may in some way be related to this finding. The

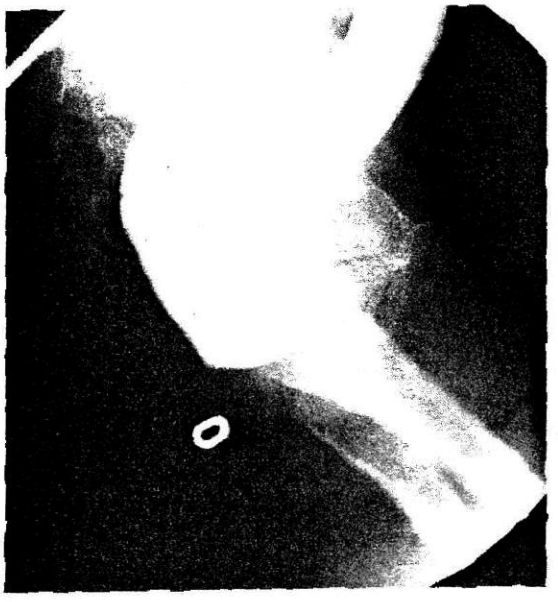

Fig 2. Distal colostogram of a patient with imperforate anus without fistula.

importance of this observation can be ascertained only when the complete embryogenesis of anorectal malformations is defined.

We have been unable to find reports on the functional results in patients with anorectal malformations and Down's syndrome. However, a tendency exists to believe that children with mental retardation and anorectal malformation will suffer from incontinence regardless of the specific type of defect and that a permanent colostomy is indicated to ease management. We were impressed that the results associated with bowel control in our patients with Down's syndrome were not different from those in the control group. Our results support the fact that patients with Down's syndrome and anorectal malformations have a similar likelihood to achieve continence as patients without Down's syndrome. This may not necessarily be true for patients with other forms of mental retardation.

Constipation is anticipated in patients with Down's syndrome alone[18] and in patients with imperforate anus without fistula.[1] We expected that when these features were combined, constipation would be worse, but we were unable to demonstrate this.

The rectal pouch in the patients without fistula lies at the level at which rectourethral bulbar fistula occurs, and the functional results in patients without fistula and in those with rectourethral bulbar fistula are similar.[11] This suggests that the height of the anorectal defect, and not the presence or absence of fistula, is the major determinant of continence.

REFERENCES

1. Black CT, Sherman JO: The association of low imperforate anus and Down's syndrome. J Pediatr Surg 24:92-94, 1989
2. Levy J: The gastrointestinal tract in Down syndrome. Prog Clin Biol Res 373:245-256, 1991
3. Zlotogora J, Abu-Dalu K, Lernau O, et al: Anorectal malformations and Down syndrome. Am J Med Genet 34:330-331, 1989
4. Buchin PJ, Levy JS, Schullinger JN: Down's syndrome and the gastrointestinal tract. J Clin Gastroenterol 8:111-114, 1986
5. Urioste M, Martinez-Friaz ML: Anorectal anomalies and Down syndrome [letter]. Am J Med Genet 39:493, 1991
6. Torfs CP, Bateson TF, Curry CJR: Anorectal and esophageal anomalies with Down syndrome [letter]. Am J Med Genet 44:847, 1992
7. Joseph VT, Chan KY, Siew HF: Anorectal malformations and their associated anomalies. Ann Acad Med Singapore 14:622-625, 1985
8. Knox GE, Ten Bensel RW: Gastrointestinal malformations in Down's syndrome. Minn Med 55:542-544, 1972
9. Gross R: The Surgery of Infancy and Childhood, Its Principles and Techniques. Philadelphia, PA, WB Saunders Co, 1953, p 356
10. Al-awadi SA, Farag TI, Naguib KK, et al: Intestinal malformations and Down's syndrome in Arabs. J R Soc Med 80:61-62, 1987 (letter)
11. Peña A: Anorectal malformations. Semin Pediatr Surg 4:35-47, 1995
12. Peña A: Atlas of Surgical Management of Anorectal Malformations. New York, NY, Springer-Verlag, 1990, p 47
13. Gross GW, Wolfson PJ, Peña A: Augmented-pressure colostogram in imperforate anus with fistula. Pediatr Radiol 21:560-562, 1991
14. Martin GR, Rosenbaum KM, Sardegna KM: Prevalence of heart disease in trisomy 21: An unbiased population. Pediatr Res 25:77A, #449, 1989 (abstract)
15. Sondheimer HM, Byrum CJ, Blackman MS: Unequal cardiac care for children with Down's syndrome. Am J Dis Child 139:68-70, 1985
16. Schneider DS, Zahka KG, Clark EB, et al: Patterns of cardiac care in infants with Down syndrome. Am J Dis Child 143:363-365, 1989
17. Rich MA, Brock WA, Peña A: Spectrum of genitourinary malformations in patients with imperforate anus. Pediatr Surg Int 3:110-113, 1988
18. Cooley WC, Graham JM, Jr: Common syndromes and management issues for primary care physicians: Down syndrome—an update and review for the primary pediatrician. Clin Pediatr 30:233-253, 1991

Discussion

C.N. Paidas: We know that Down's syndrome is a chromosomal abnormality. In this paradigm of imperforate anus without fistula in association with Down's syndrome, can that paradigm be applied to the 15 or 20 additional chromosomal abnormalities that have imperforate anus as one of its associated anomalies? In other words, could you go through your 900 or so patients and extract this same sort of paradigm with other chromosomal abnormalities?

M.A. Levitt (response): That is a good question and definitely needs to be studied. As we said, many of these chromosomal anomalies seem to correlate with not only imperforate anus, but with the specific diagnosis of imperforate anus without fistula. I'm not sure why.