The Association of Low Imperforate Anus and Down's Syndrome

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● **Down's syndrome is the most frequent chromosomal anomaly in humans and is associated with an incidence of anorectal anomalies many times greater than that found among the general population. The anorectal malformation associated with Down's syndrome uniformly consists of a low-lying rectal pouch without a genitourinary or perineal fistula. This type of imperforate anus may often be adequately treated by simple perineal anoplasty. Since our recognition of this association, several neonates have avoided temporary fecal diversion, and several older infants with colostomies have not required anticipated pull-through procedures.**

INDEX WORDS: Anorectal anomalies; Down's syndrome.

MORE THAN 2% of all children born with an anorectal anomaly also have Down's syndrome. This is an incidence 15 times more frequent than the rate of 0.15% found in the general population.[1-7] Because of this apparent association, we sought to determine if consistent features characterize anorectal anomalies occurring in conjunction with Down's syndrome.

MATERIALS AND METHODS

Fourteen children with Down's syndrome and an imperforate anus treated over the past 10 years were studied. There were eight females and six males. Three patients were neonates. Four patients were infants from 1 to 12 months of age, each of whom had been diagnosed at birth as having a high imperforate anus. Although colostomies had been created for each shortly after birth, no pull-through procedures had yet been performed. Seven former patients, treated initially by colostomy and subsequently by a pull-through operation, were also evaluated through a review of their hospital records.

Following the diagnosis of imperforate anus, a physical examination of each child's perineum for fistulae was performed. The bladder and urethra were examined for fistulae with voiding cystourethragraphy (VCUG). The kidneys and ureters were evaluated for anomalies in each case by intravenous pyelography (IVP) or by ultrasonography (US).

Soon after the birth of each child, the level of the rectal pouch or the pouch-to-perineum (P-P) distance was evaluated. Three of the 14 children were assessed solely on the basis of the physical examination. The invertogram was the only other test performed in eight others. US of the perineum and pelvic contents was employed in the three remaining newborns, although invertograms of two of these patients were also obtained.

Four infants underwent additional more-detailed examinations after colostomies were constructed. The pouch level for each of these children was further evaluated by US (three patients), by distal contrast stomatography (DCS; three patients), or by passage of a catheter through the distal stoma into the blind-ending rectum (one patient). Direct measurements of the P-P distance were recorded at the time of surgery for six of the 14 patients.

RESULTS

Results are summarized in Table 1. The P-P distances of six children ranged from 5 to 10 mm at the time of surgery (patients 1 through 6). Five of the six children had undergone preoperative evaluation by perineal US, which had shown low-lying pouches in four; the US study of the fifth was of poor quality. After patient 7 underwent a colostomy, a catheter was passed through the distal stoma into the rectal pouch. The tip of the catheter caused the perineum to bulge at the anal dimple, and the P-P distance was estimated (by palpation of the catheter tip) to be not >10 mm. Patient 8 had a P-P distance of 14 mm determined by US, but no measurements were taken at the time of the operation. The invertograms of two other neonates (patients 9 and 10) showed gas patterns below the pubococcygeal (P-C) line. Patient 11 received a colostomy at birth, but at 15 months of age was found to have a pinhole-sized opening at the site of the normal anus. This anal stenosis was dilated to a functional anus prior to colostomy closure. The rectal pouches of patients 12 and 13 were visualized at the I-point by DCS. Although contrast extending no further than the I-point on DCS is believed to indicate an intermediate-level pouch, such studies were obtained in patients 5 and 6, who were later found to have low pouches. The rectal pouch of patient 14 was found to extend so far through the levator muscles at the time of the laparotomy that the intended pull-through procedure was abandoned in favor of a perineal anoplasty which was easily accomplished.

Neither genitourinary fistulae nor renal or ureteral anomalies were found in any of these 14 children. One

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Table 1. Data for 14 Patients With Down's Syndrome and Imperforate Anus

Patient No./Sex	US or IVP of Kidneys and Ureters	Bladder and Urethra	Fistula	Extent of Rectal Pouch		
				Operative Report	Invertogram (*) Stomatogram (#)	Perineal US
1/F	US NL	NL	None	P-P = 10 mm	*Above P-C line	P-P = 14 mm
2/M	US NL	NL	None	P-P = 6 mm	Neither performed	P-P = 9 mm
3/F	US NL	NL	None	P-P = 8 mm	*Above P-C line #Below I-point	P-P = 8 mm
4/F	US NL	Long urethra	None	P-P = 5 mm	*Below P-C line	P-P = 6 mm
5/F	US NL	NL	None	P-P = 6 mm	*Above P-C line #At I-point	P-P < 20 mm
6/M	IVP NL	NL	None	P-P = 10 mm	*Below P-C line #At I-point	Not performed
7/F	US NL	NL	None	Estimated < 10 mm	Neither performed	Not performed
8/M	US NL	NL	None	Not stated	Neither performed	P-P = 14 mm
9/M	IVP NL	NL	None	Not stated	*Below P-C line #Above P-C line	Not performed
10/F	US NL	NL	None	Not stated	*Below P-C line #Above P-C line	Not performed
11/F	US NL	NL	Anal stenosis	No operation	*Above P-C line	Not performed
12/M	IVP NL	NL	None	Through levators	*Above P-C line #At I-point	Not performed
13/F	US NL	NL	None	Not stated	#At I-point	Not performed
14/M	IVP NL	NL	None	Through levators	*Above P-C line	Not performed

Abbreviation: NL, normal.

infant girl was found, incidentally, to have an unusually long urethra upon VCUG, but has normal function.

DISCUSSION

An important, but previously unrecognized, association exists between Down's syndrome and low imperforate anus without a fistula. This malformation consists of an incomplete development of the proctodeal pit. Anomalies resulting from such a developmental failure include anal agenesis without fistula, anorectal stenosis, covered anal stenosis, imperforate anal membrane, and anal membrane stenosis. Any or all of these conditions may have been represented by patients in the present series. Although embryologically each of the deformities is anal in origin, Stephens and Smith[2] consider the first two as intermediate and the others as low.

Eighteen years ago, a survey of the Surgical Section of the American Academy of Pediatrics (AAP) reported the frequency with which various types of anorectal anomalies are seen. A "low" anomaly in that series was defined as "translevator," and those anomalies comprised 48% of the total. All 14 children in the present series had translevator or "low" anomalies. Although 38% of the children in the AAP series had an upper urinary tract anomaly, none was seen in our group. Forty-nine percent of the children in the AAP series had a urogenital fistula, while none was found in these children with Down's syndrome.[8] Stephens and Smith[2] state that more than 93% of females and 86% of males with anal anomalies will have an external or urogenital fistula. In our group, 12 might have been expected to have fistulae, but none did.

Eleven of our patients had been treated initially by creation of a diverting colostomy. Such treatment was based on erroneous interpretations of physical and radiographic examinations, but is in accordance with currently accepted principles of management. A perineal anoplasty may be appropriate treatment for selected newborns with Down's syndrome and an imperforate anus.

This association has gone unnoticed because of the inaccuracies inherent in the present methods of pouch level evaluation. It is likely that the true incidence of low imperforate anus occurs more frequently than presently believed, due to the tendency of current tests to mistake lower for high anomalies but seldom the reverse. If this is true, and these neonates are identifiable, perineal anoplasty in the newborn could be performed more frequently, and some pull-through procedures could be avoided.

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Discussion

F. Guttman (Montreal): I am not sure that this report is telling us something new. My first patient with this problem dates from the early 70s when Brad Rodgers was a resident in Montreal. Since then, I and many others have seen both high and low types associated with Down's syndrome. Down's syndrome as an early embryological event is often associated with other major defects—cardiac, duodenal, and anorectal. In a large survey of anorectal anomalies carried out by the French Pediatric Surgical Society last year, there were many low and high types associated with Down's syndrome. You have a selected experience. Perhaps your next 14 patients will have Down's syndrome with high-type association.

C.T. Black (closing): Dr Guttman, I appreciate your comments. We do not know of any reason to assume that the presence of Down's syndrome precludes the random occurrence of other types of anorectal anomalies. In fact, given the increased frequency of anorectal anomalies in Down's syndrome, I would expect that about one out of every 15 or so children who have both conditions would have a "nonassociated" anomaly. In such a situation, any of the usual spectrum of variations might be observed. This presentation was not meant to suggest that such an occurrence does not exist. Instead, we wish to draw attention to an apparent frequent association, and to encourage particularly accurate evaluations of pouch levels in these children. We have shown that the anorectal anomaly may very often be easily corrected in the newborn period without a colostomy or later reconstructive surgery.