Presentation of Low Anorectal Malformations Beyond the Neonatal Period

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ABSTRACT. Objective. Anorectal malformations are usually diagnosed at birth, but some patients have presented to this institution beyond the early newborn period without recognition of their anorectal malformations. To quantify the extent of this problem, we undertook a review of all patients presenting to this hospital with anorectal malformations.

Methods. We reviewed all new cases of anorectal malformations treated at British Columbia’s Children’s Hospital during the past 11 years. We looked specifically at the time of diagnosis, patient age, sex and mode of presentation, the type of anorectal malformations, and any associated anomalies.

Results. One hundred twenty new cases of anorectal malformations were seen here, of whom, 15 patients (9 girls and 6 boys) presented beyond the early newborn period. Of these, 1 male infant was diagnosed at 2 weeks of age and another girl at 14 years of age. The remaining 13 presented between 3 and 11 months of age because of increasing constipation, usually associated with the introduction of solid foods. All had low anorectal malformations. Nine patients had at least 1 other feature of the VACTERL complex.

Conclusions. Most anorectal malformations are identified at birth, but a significant number of the milder lesions may not be recognized until later. Therefore, this condition must be considered in older infants and children presenting with constipation, particularly if they also have cardiac or genitourinary anomalies.

ABBREVIATION. VACTERL, vertebral abnormalities, anal atresia, cardiac abnormalities, tracheoesophageal fistula and/or esophageal atresia, renal agenesis and dysplasia, and limb defects.

Anorectal malformations are a group of congenital conditions comprising a spectrum in severity from imperforate anal membrane to complete caudal regression. The incidence of such abnormalities is ~1 of every 5000 newborns.1 We have noted at our institution that occasionally patients with congenital anorectal malformations have not had this condition diagnosed during the neonatal period. Such a delayed diagnosis may complicate the surgical repair and may contribute to both functional and psychological problems for the patient and family. Previous studies reported in the literature have been limited to few case reports.2,3

To provide a measure of the rate of missed anorectal malformations in the newborn period, we have reviewed all new cases of anorectal malformation presenting to our institution over the past 11 years. We looked specifically at the age and mode of presentation. Also noted were patient sex, type of anorectal malformation, and the presence of associated anomalies.

METHODS

All cases of patients newly presenting with anorectal malformations over an 11-year span from January 1987 to November 1997 at the British Columbia’s Children’s Hospital were identified from the medical records. The British Columbia’s Children’s Hospital is the only tertiary pediatric referral center for the province of British Columbia, serving a population of over 3 million people. For this study, we defined a late diagnosis of anorectal malformation as a case in which a newborn patient was discharged from hospital without recognition of the anorectal malformation or referral to a pediatric surgeon. Specific details about patient characteristics, the pattern of presentation, and treatment were obtained from the hospital charts and surgeons’ records.

RESULTS

Over the 11-year period of study, 120 new cases of anorectal malformation (58 girls and 62 boys) presented to the British Columbia’s Children’s Hospital. In 15 patients (13%), the anorectal malformation was not identified in the early newborn period. There were 9 girls (Table 1) and 6 boys (Table 2) in this group. Thirteen of these 15 patients presented between 3 and 11 months of age, evenly distributed throughout this period. All but 1 of the children in this group presented with constipation, consistently coinciding with a change in their diets from breast milk to formula or at the time of introduction of solid foods. Two patients also were noted to pass very small caliber stools. Several of the patients had been seen by more than 1 physician, some of whom did not examine the child’s perineum. The 2 patients in this group not presenting within this 3- to 11-month age range included a 2-week-old male infant who was identified when his parents noticed urine draining from his anus (Table 2, case 10), and a 14-year-old girl who for many years had passed only 1 bowel movement every 2 to 3 weeks (Table 1, case 9). In the former case, this male infant was found to have a duplicate urethra with 1 tract draining into the perineal fistula.

Careful physical examination confirmed the diagnosis in all the cases. Typically the perineum was

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raw and irritated. In boys, visual inspection revealed a distinctly abnormal orifice (Fig 1). The opening was anterior to the external sphincter complex and the margins lacked the usual radial corrugations seen at the anus. By several months of age, this was likely more apparent than at the time of birth. The distal tract was densely fibrotic and was too narrow to admit an examining finger. The appearance was similar in girls with perineal fistulas, whereas in those girls with vestibular fistulas, the orifice was anterior to the posterior fourchette of the vagina.

All 15 patients had low anorectal malformations; all boys had a perineal fistula (Fig 1), while girls had either a rectovestibular fistula (8) or a perineal fistula (1). In addition, 9 children had at least 1 other feature of the VACTERL complex, including the urinary system (5), the heart (3), the spine (3), and the upper limb (2; Tables 1 and 2). All patients but 1 were treated initially with a diverting colostomy and subsequent posterior sagittal anorectoplasty; the 1 exception had a primary anorectoplasty. In several cases, including the patient who presented at 14 years of age (Table 1, case 9), correction was surgically challenging because of the massive colonic dilatation resulting from the chronic obstruction. After the anorectoplasties, the colostomies have been closed in 13 patients; 1 patient had a primary anorectoplasty and the other presented toward the end of the study.

DISCUSSION

In this review of our recent experience, 15 children with delayed recognition of their anorectal malformations were identified. Within this group, consistent findings were observed in terms of clinical presentation and age at diagnosis. It seems that most children suffered from chronic constipation that became worse and/or refractory to medical treatment around the time of dietary change from breast milk to formula or at the time of introduction of solid foods; others became frankly obstipated. This reflects the greater difficulty of passing the more solid stool through the abnormal distal channel. All but 2 children in this group presented in this way between the ages of 3 and 11 months. This observation is important because it has not been reported previously in a group of patients; indeed, it has been stated that conservative treatment of anterior ectopic anus is generally adequate.

As anticipated, the missed anorectal malfor-
tions were all the low type. In addition, the type of malformations found in girls and boys were consistently of the same type for the respective sexes. There was a slight preponderance of girls in this group (9 girls vs 6 boys), compared with the overall higher frequency of anorectal malformations in boys seen in this and other series. This may reflect the relatively higher frequency of low anorectal malformations in girls, because these are the lesions likely to be missed in neonates. It is difficult to be definitive about distinguishing a normal from an abnormal anus. In general the abnormal anus will be located more anteriorly and sometimes lateral to the midline (Fig 1). It will also tend to lack the radial corrugations produced by the underlying anal sphincter. However, in the older infant or child, it is important to correlate the appearance of the anus with its function.

In all but 1 case, a divided colostomy was constructed before performing the anoplasty. This was done because in most cases the perineum had become raw and inflamed from the continual soiling. The colostomy allowed the perineum to heal before the anoplasty and protected the surgical repair postoperatively. Also, in several cases the colon had become markedly dilated from the partial obstruction, so the colostomy was necessary to provide adequate decompression.

More than half of the children in this series had at least 1 other feature of the VACTERL complex; anomalies of the urinary system, which are commonly associated with anorectal malformations, were found in 5 children. Therefore, the index of suspicion of an associated anorectal malformation should increase if a child presents with chronic constipation and any feature(s) of genitourinary, cardiac, spine, or limb anomalies. Careful inspection of the perineum and digital rectal examination should be performed on any infant or child being evaluated for constipation.

This study highlights some important features of children with delayed diagnosis of anorectal malformation. The delayed diagnosis of the anorectal malformation has the potential to contribute to unnecessary patient suffering and in our experience diminished parental confidence in their physicians. It is important to be aware that not all anorectal malformations will present in the newborn period as an imperforate anus. Examination of the anal region in the newborn should involve more than just confirming the presence of an anal orifice. Attention should be taken to note the location and appearance of the anus. Clinical suspicion should be heightened in infants or toddlers who present with an exacerbation of constipation associated with a recent change in diet, especially if they have 1 or more features of the VACTERL complex. Any suspicion of an abnormally located anal opening, regardless of its subtlety, warrants referral to a surgeon with expertise in managing anorectal malformations.

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