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Screening practices and associated anomalies in infants with anorectal malformations: Results from the Midwest Pediatric Surgery Consortium☆

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ABSTRACT

Background: This study evaluates screening practices and the incidence of associated anomalies in infants with anorectal malformations (ARM).
Methods: We performed a multi-institutional retrospective cohort study of children born between 2007 and 2011 who underwent surgery for ARM at 10 children's hospitals. ARM type was classified based on the location of the distal rectum, and all screening studies were reviewed.
Results: Among 506 patients, the most common ARM subtypes were perineal fistula (40.7%), no fistula (11.5%), and vestibular fistula (10.1%). At least 1 screening test was performed in 96.6% of patients, and 11.3% of patients underwent all. The proportion of patients with ≥1 abnormal finding on any screening test varied by type of ARM ($p < 0.001$). Screening rates varied from 15.2% for limb anomalies to 89.7% for renal anomalies. The most commonly identified anomalies by screening category were: spinal: tethered cord (20.6%); vertebral: sacral dysplasia/hemisacrum (17.8%); cardiac: patent foramen ovale (58.0%); renal: hydronephrosis (22.7%); limb: absent radius (7.9%).
Conclusion: Screening practices and the incidence of associated anomalies varied by type of ARM. The rate of identifying at least one associated anomaly was high across all ARM subtypes. Screening for associated anomalies should be considered standard of care for all ARM patients.
Type of study: Multi-institutional retrospective cohort study.
Level of evidence: III

Anorectal malformations (ARMs) are rare congenital anomalies of the distal gastrointestinal and urogenital tracts that occur with an incidence of approximately 1 in 5000 live births [1]. ARMs exist on a spectrum of severity, from relatively simple anal anomalies to complex defects. While ARMs may exist as an isolated defect, they are known to be associated with various multisystem anomalies in the vertebral,

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anorectal, cardiac, tracheoesophageal fistula, renal, and limb (VACTERL) syndrome [2–4]. The rate of associated anomalies varies by the complexity of ARM, with more severe malformations demonstrating a higher rate of associated anomalies compared to the less severe variants [1,5,6].

Despite the well-known and frequent associations with other anomalies, screening rates for these associations have been shown to be low [7,8]. This may be because of a variety of reasons, including a belief among some practitioners that patients with less severe malformations do not require a full complement of screening studies [9]. Beyond identifying anomalies that are of immediate concern in the newborn period, screening in this population permits the diagnosis of disease that may not become clinically significant until later in life. Thus, failure to screen for and identify associated anomalies may lead to increased morbidity both in early childhood and later in life [10–16].

Literature on the rates of screening and identification of associated anomalies in patients with ARMs is limited. The objective of this study was to identify the screening rates and rates of associated anomalies in patients with ARMs across 10 children's hospitals and to determine the rate of associated anomalies detected by type of ARM.

1. Methods

1.1. Study design and patient population

We performed a multi-institutional retrospective cohort study of patients born between January 2007 and December 2011 who underwent primary corrective surgery for an ARM at 10 children's hospitals participating in the Midwest Pediatric Surgery Consortium (www.mwpsc.org). Patients were identified for inclusion based on International Classification of Disease, 9th Edition, Clinical Modification (ICD-9-CM) diagnosis codes for ARM (565.1, 596.1, 619.1, 599.1, 602.8, 751.2, 751.5, 752.49, 753.8). At each institution, the medical records of all patients with one of these diagnosis codes were reviewed to verify the presence of an ARM. The 2007 to 2011 time period was chosen because the primary objective of the project is the development of a predictive index for fecal incontinence in children with ARMs, thus we included only children 4 years of age or older at the time of the retrospective chart review. Patients included in this investigation are currently being followed up prospectively for the assessment of continence, and these findings will be reported in a future publication.

1.2. Data collection

Patient demographics and clinical characteristics were extracted from the medical record. In cases where a patient was referred from an outside hospital to one of the participating institutions for definitive repair, all records available from the outside institution were reviewed and identified tests performed and their results were included. Each patient's type of ARM was classified based on the location of the distal rectum from imaging and operative reports [17]. We also collected data on whether screening tests for specific organ systems were performed, and we noted the findings from those tests. Screening tests evaluated included spine radiographs for vertebral anomalies, lumbosacral radiographs for sacral anomalies, spinal ultrasound or MRI for spinal cord anomalies, echocardiogram for cardiac anomalies, renal or abdominal ultrasound for renal anomalies, and limb radiographs or ultrasounds for limb anomalies. Any of these tests performed within the first year of life were considered for screening purposes and included. For the purpose of this study, we excluded evaluation for esophageal atresia with or without tracheoesophageal fistula (EA/TEF), as routine screening for EA/TEF is essentially performed in every newborn with the initiation of feeding. If a newborn does not tolerate feeds, then a nasogastric tube is passed, and failure of passage may identify EA/TEF. Therefore, EA/TEF was excluded from this analysis in order to focus on the screening for anomalies that are not a mandatory part of routine perinatal care.

1.3. Statistical analyses

Frequencies and percentages were used to describe categorical variables, and medians and interquartile ranges were used to describe continuous variables. As appropriate, either Pearson chi square or Fisher exact tests were performed to compare rates of screening and rates of abnormal findings on screening tests by type of ARM. In analyses of screening rates, all patients in the cohort were included. In analyses of the rate of abnormal findings on screening tests, only included patients that had undergone a screening test for that system(s) were included. Patients who had a record of having undergone a screening test but with no result available were included in the analysis of the screening rates but not the rates of associated anomalies. A P value <0.05 was considered statistically significant. All analyses were conducted using SAS 9.4 (SAS Institute Inc., Cary, NC). This study was approved by the Institutional Review Board at all participating institutions.

2. Results

A total of 506 patients were included. Demographic and clinical characteristics of the cohort are shown in Table 1. The majority of patients were white, there was an even gender distribution, and the majority of patients were under 1 year of age at the time of their corrective surgery (93%). The most common ARM subtypes were perineal fistula (40.7%), rectal atresia with no fistula (11.5%), and vestibular fistula (10.1%).

The rates of associated anomaly screening by organ system are shown in Table 2. The renal system was the most common system evaluated with 89.7% patients undergoing renal ultrasound. Vertebral

Table 1
Cohort demographics and clinical characteristics.

	Number of patients (%)
Sex	
Male	251 (49.6)
Female	255 (50.4)
Race	
White	354 (70.1)
Black/African American	53 (10.5)
Asian	16 (3.2)
Native Hawaiian or Other Pacific Islander	2 (0.4)
American Indian or Alaskan Native	1 (0.2)
Biracial or Multiracial	14 (2.8)
Unknown	65 (12.9)
Number of Patients per Institution	
Hospital 1	92 (18.2)
Hospital 2	90 (17.8)
Hospital 3	83 (16.4)
Hospital 4	64 (12.6)
Hospital 5	47 (9.3)
Hospital 6	39 (7.7)
Hospital 7	29 (5.7)
Hospital 8	22 (4.3)
Hospital 9	21 (4.2)
Hospital 10	19 (3.8)
Type of ARM	
Perineal fistula	206 (40.7)
Vestibular fistula	51 (10.1)
Rectobulbar fistula	27 (5.3)
Rectovesical fistula	23 (4.5)
Rectovaginal fistula	21 (4.2)
Rectoprostatic fistula	22 (4.3)
Cloaca <3 cm common channel	18 (3.6)
Cloaca >3 cm common channel	31 (6.1)
No fistula	58 (11.5)
Pouch colon	1 (0.2)
Rectal stenosis, anal stenosis	16 (3.2)
High imperforate anus	4 (0.8)
Imperforate anus	4 (0.8)
Cloacal extrophy	20 (4.0)
Anal atresia	2 (0.4)
H fistula	2 (0.4)