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Lack of Disparities in Screening for Associated Anomalies in Children with Anorectal Malformations

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Abstract

INTRODUCTION—Patients with anorectal malformations (ARM) often have associated congenital anomalies and should undergo several screening exams in the first year of life. We hypothesized that racial and socioeconomic disparities exist in the screening processes for these patients.

METHODS—Following IRB approval, a retrospective review of patients with ARM born between 2005 and 2016 was performed at a quaternary care children's hospital. Demographics including gender, race, insurance, and zip code were collected. Zip code was used as a surrogate for median income. Chart review was performed to identify anomaly type and whether VACTERL screening was performed within one year of age. Descriptive statistics and Chi square analyses were performed.

RESULTS—100 patients (59% male, 68% low malformation) were identified. African-American and Caucasian subjects represented 41% and 40% of the population, respectively. Overall, 68/100 patients had at least one screening test for each of the VACTERL associations. Although some minor differences were noted (more African-Americans received skeletal survey than Caucasians, 80.5% versus 60%, $p=0.00335$), no pattern of systematic bias in the receipt or timing of screening was evident based on race, insurance, or income.

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CONCLUSIONS—There do not appear to be racial or socioeconomic disparities in screening for associated anomalies in patients with ARM. However, overall gaps in screening still exist, and work must be done to appropriately screen all patients for associated anomalies.

Keywords

Anorectal malformations; congenital anomalies; VACTERL; disparities; neonatal

Introduction

Anorectal malformations (ARM) are a group of rare congenital defects that result from abnormal development of the hindgut, allantois, and Mullerian ducts. ARM have been reported to occur in approximately 1 out of every 4000–5000 live births, with a slight male predominance [1]. While ARM represent a rare category of congenital defect, they are frequently associated with other defects. The VACTERL association was first described in 1973 as the non-random association of vertebral, anorectal, cardiac, tracheoesophageal fistula/esophageal atresia (TEF/EA), renal, and limb anomalies [2]. Patients are classified as exhibiting the VACTERL association if they are diagnosed with 3 or more of these associated anomalies. The rate of VACTERL associated anomalies in ARM patients has been reported as high as 71% [3][4][5][10]. High lesions have been demonstrated to have a higher incidence of associated anomalies compared to low lesions [10].

The presence of undiagnosed anomalies in ARM patients is a potential source of morbidity and mortality that extends well into adulthood, emphasizing the importance of screening protocols [8][9]. Several screening recommendations have been suggested, including screening for other anomalies in patients with two VACTERL anomalies, screening all patients with TEF/EA and ARM, and screening all ARM for tethered cord [6][14]. Implementation of screening protocols has been demonstrated to improve anomaly detection in ARM patients, although questions still remain about ideal screening modality and timing [7].

Socioeconomic status (SES) is recognized as an important determinant of pediatric health outcomes. Race and income impact both health status and utilization of health services, with lower SES children less frequently utilizing health resources [11][15]. Lower SES has been associated with worse outcomes in children in a wide variety of health parameters [16]. Previous studies have demonstrated that children with public insurance were less likely to receive specialty care than those with private insurance [12][13][17][18]. We hypothesized that lower SES ARM patients received less VACTERL screening.

Materials and Methods

The University of Tennessee Health Science Center Institutional Review Board approved this study and, in accordance with the Code of Federal Regulations Title 45 (part 46, subpart D), granted waiver of informed consent. A single-institutional, retrospective cohort study was performed for patients born between 2005–2016 at an urban, quaternary care, free-standing children's hospital. Patients with ARM were identified using International Classification of Diseases (ICD) 9 code 751.2 and ICD-10 codes Q42.3 and Q43.6.

Diagnoses were then confirmed by individual electronic medical record review. Patients were excluded if the diagnosis was not documented in the medical record, if surgery was performed prior to 2005 (when the electronic medical record was implemented), or if there was no documentation of screening studies in patients who were transferred from other institutions. Patients with cloaca were also excluded. Demographics including gender, race, insurance, and zip code were extracted from the medical record. Zip code of residence was used as a surrogate for median income based on currently available census data [19].

Chart review was performed to specify ARM anomaly type and to identify receipt and timing of VACTERL screening studies, including chest x-ray, skeletal survey, “babygram”, renal ultrasound, echocardiogram, spine ultrasound, and spine MRI. We used skeletal survey and babygram for vertebral anomaly screening, chest X-ray for TEF screening, echocardiogram for cardiac screening, renal ultrasound for renal anomalies, skeletal survey for limb anomalies, and spine ultrasound and MRI for spinal anomaly screening. ARM were grouped into “low” or “high” using modifications of the Wingspread and Krickenbeck classification systems. Low malformations include anal stenosis and perineal and vestibular fistula. High malformations include rectourethral and rectovesicular fistulas.

Demographics and screening tests are reported with descriptive statistics and Chi square analysis were performed. Non-parametric tests were used where appropriate. A p-value <0.05 was considered statistically significant. All analyses were conducted in SAS® 9.3 (Cary, NC).

Results

Patient Demographics

We identified 101 patients (59% male, 68% low malformation) during the study period (Table 1). One patient died on day of life 1 and was excluded from analysis. African-American and Caucasian subjects represented 41% and 40% of the population, respectively. The majority of subjects (69%) had government insurance. Median incomes ranged from \$23,355-\$142,928, with the 50th percentile income level at \$36,857.

Overall, 87 patients (87%) had at least one associated anomaly and 28 patients (28%) met the definition for VACTERL association (≥ 3 anomalies). Of those with VACTERL, 13 patients (46.4%) had low anomalies and 11 patients (39.3%) had high anomalies. There were no differences in race, insurance status or income among those patients with additional anomalies or VACTERL association.

Receipt of Screening

An imaging study qualified as a screening test if it was performed within the first year of life. All documented images were used, regardless of the indication for the study. Overall, 68 of 100 patients (68% Table 2) had at least one screening test for each of the VACTERL associations. There were no differences in screening rates by race (Table 2). With respect to insurance, 100% of patients with commercial insurance received spinal screening versus 75% of patients with private insurance and 72% of patients with government insurance ($p=0.0188$). Neither of the 2 patients in the lowest income bracket received limb screening

compared to 85% in the 25–35k group, 68% of the 35–50k group, and 68% in the 50k+ group ($p=0.0453$). None of the screening types were found to be completed in 100% of the patients during the study period (Table 3). Renal ultrasound and echocardiogram each had an 89% completion rate. Spine MRI had the lowest rate of completion (25%). Of note, all spine ultrasounds were completed before 6 months of age. However, 20 (20%) patients had neither a spine ultrasound nor an MRI documented, while 28 (28%) had both studies. Out of 28 patients who had both a spine ultrasound and MRI, 3 of them had an abnormal MRI result after having a normal ultrasound (10.7%). The median number of studies per patient was 5 (IQR 4–6). There was no difference in median number of studies when comparing by race, insurance, or income group. Patients with high anomalies received a greater number of studies (high anomalies = 6, IQR 5–6; low = 5, IQR 4–6; $p=0.0368$).

Time to Screening and Results of Screening

Of those screenings that were completed within one year of birth, the most common to show abnormal results were echocardiograms (89.9%), spine MRI (52.5%), spine ultrasound (40.3%), and skeletal surveys (37.5%, Table 3).

With respect to the time from birth to completion of a study (including those performed after one year of life), there were some significant differences noted when comparing different insurance types and income groups, however no pattern of systematic bias was evident (Figure 1). For the subset of patients with abnormal study results, there were no differences in overall time to study completion by race or income; however, subjects with government insurance and abnormal spine ultrasounds had a longer median time to ultrasound than subjects with other insurance types (Government = 7 days, IQR 5–21; Private = 2.5, IQR 0.7–4; Commercial = 1.7, IQR 0.5–2; $p=0.014$).

Discussion

Thorough evaluation for VACTERL associations in children with ARM is critical to providing appropriate and timely care. Our review of 100 patients with ARM at an urban, quaternary care free-standing children's hospital found suboptimal rates of screening for VACTERL association. Only 68% of patients received at least one screening test for each type of associated anomaly. Cardiac, renal, and TEF/EA anomalies had the highest screening rates, while spinal and limb anomalies had the worst screening rates. There were disparities noted in terms of median time to skeletal surveys, but there was no evidence of systemic disparities in our institution, which has an equal distribution of patients in terms of race and broad representation of socioeconomic statuses. These data represent an important foundation for future study involving the screening of patients with ARM.

Overall screening rates for VACTERL association are lacking in modern literature. The largest recent study examined 406 patients with ARM from the Medicaid Analytic eXtract database who had a definitive ARM surgical procedure within the first year of life [20]. They found an overall screening rate of 13%. In this population, vertebral anomalies had the lowest screening rate (8%), and genitourinary anomalies had the highest screening rate (84%). In contrast, we found a significantly higher rate of screening (68%), which may be reflective of increased awareness of the need to screen for associated anomalies when these

neonates are treated at children's hospitals. Of note, the Medicaid study did not specify the time period after birth in which the screening tests are performed. We believe that early screening has important implications for the management of children with ARM. To illustrate, lipomyelomeningocele, which is the most common form of occult spinal dysraphism, often presents with bladder dysfunction in the first 2 years of life, followed by motor or sensory symptoms in later years [21]. Patients with this defect are more likely to have symptoms after 6 months of age rather than in the first 6 months of life [21]. This highlights the need for spinal screening in ARM in the first year of life.

Screening for VACTERL association is of paramount importance in patients with ARM. The associated anomalies may contribute significantly to a patient's functional status and short- and long-term quality of life, in addition to any ongoing problems they may have from a gastrointestinal standpoint. A recent study compared surgical and clinical outcomes in ARM children with and without VACTERL association [22]. Their patient population had an incidence of 31% ARM with VACTERL, and 66% of ARM patients had at least one associated anomaly. They found that children with VACTERL association had significantly higher rates of surgical complications (29% vs 10.5%, $p=0.0174$) as well as increased need for bowel management (54.8% vs 32.9%, $p=0.0251$) and dietary/laxative treatment (93.5% vs 56%, $p<0.0001$). They suggested that the poorer clinical outcome may be related to the higher incidence of spinal anomalies in VACTERL patients, as this relationship has been demonstrated previously [23].

Patients with low ARM anomalies, as compared to high, are thought to have a lower incidence of associated anomalies [20], and there may be a temptation to perform less screening tests in patients with rectoperineal fistula, which is the mildest variant of ARM. In our study, we did see that patients with higher ARM anomalies underwent more screening tests compared to those with low anomalies. However, one group studied 308 patients with rectoperineal fistula and reported that 34% were found to have at least one associated anomaly, despite incomplete screening in a third of patients [24]. Therefore, these patients still require full screening.

Our study has some limitations. These findings are specific to a single quaternary care center with a heterogeneous patient population. However, our institution has an equal mix of African-American and Caucasian patients and a broad socioeconomic catchment area, which may be representative of other freestanding children's hospitals. Additionally, there may have been missing documentation of screening, particularly in patients who were transferred from another institution soon after birth. We consider this unlikely, as these infants are typically transferred to our institution within 24 hours of birth and most screening studies happen beyond that time period.

Conclusions

Children with ARM are at risk for associated VACTERL anomalies. We found a 68% overall screening rate at our tertiary hospital, and no evidence of systemic disparities in screening. In order to improve the care, quality of life, and outcomes of these children, clinical pathways that include appropriate screening for these children may be beneficial.

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Figure 1.

Median days to screening for VACTERL anomalies by Insurance, Race and Income. Median days (IQR) to screening for VACTERL anomalies by (A) Insurance, (B) Race, and (C) Income are presented. Vertebral = skeletal survey or babygram; Cardiac = echocardiogram; TEF/EA = chest x-ray; Renal = renal ultrasound; Limb = skeletal survey; Spinal = spine ultrasound or spine MRI. * *statistically significant result*.

Table 1

Demographics.

Gender	% (n=100)
Female	41%
Male	59%
Race	
African-American	41%
Caucasian	40%
Other	18%
Low Anomaly	68%
Anal Stenosis	3%
Perineal fistula	45%
Vestibular fistula	20%
High Anomaly	24%
Rectourethral fistula	13%
Rectovesicular fistula	3%
Rectovaginal fistula	2%
No fistula	6%
Other/Unknown Anomaly	8%
Insurance	
Government	67%
Private pay	12%
Commercial	18%
Missing	3%
Median Income	
<25k	2%
25k–35k	33%
35k–50k	41%
>50k	19%

Table 2

VACTERL screening in children with ARM during the first year of life.

Screening Test	Patients Receiving Screening	Screening by Race	Screening by Insurance	Screening by Income
Vertebral Babygram and/or Skeletal survey	82%	<i>P=0.5873</i>	<i>P=0.7236</i>	<i>P=0.3449</i>
Cardiac Echocardiogram	89%	<i>P=0.7074</i>	<i>P=0.4417</i>	<i>P=0.75</i>
TEF/EA CXR and/or Babygram	83%	<i>P=0.2705</i>	<i>P=0.1662</i>	<i>P=0.4982</i>
Renal Renal ultrasound	89%	<i>P=1</i>	<i>P=0.873</i>	<i>P=0.7243</i>
Limb Skeletal survey	72%	<i>P=0.0823</i>	<i>P=0.6436</i>	<i>P=0.0453*</i>
Spinal Spine ultrasound and/or Spine MRI	77%	<i>P=0.5505</i>	<i>P=0.0188*</i>	<i>P=0.9103</i>
At least one screening test per category	68%	<i>P=0.8185</i>	<i>P=0.9438</i>	<i>P=0.9778</i>

* indicates a significant result.

Table 3

Study Completion Rate and Rate of Abnormal Findings

Study	Studies documented in 1st year of life	Studies with abnormal findings	Most common abnormal finding (n)
Chest x-ray	82%	19.5% (16/82)	Vertebral anomaly (7)
Skeletal survey	72%	37.5% (27/72)	Vertebral anomaly (13)
Babygram	53%	20.8% (11/53)	Vertebral anomaly (6)
Renal ultrasound	89%	30% (24/89)	Hydronephrosis (12)
Echocardiogram	89%	89.9% (80/89)	Patent Foramen Ovale (53)
Spine ultrasound	72%	40.3% (29/72)	Low conus (15)
Spine MRI	25%	52.5% (13/25)	Low conus (12)