Congenital Anorectal Malformation Severity Does Not Predict Severity of Congenital Heart Defects

Jara E. Jonker, BSc[1], Eryn T. Liem, MD, PhD[2], Nynke J. Elzenga, MD, PhD[2], Bouwe Molenbuur, MD[3], Monika Trzpis, PhD[4], and Paul M. A. Broens, MD, PhD[1,4]

Objective To determine the prevalence of congenital heart defects (CHDs) in patients with mild or severe congenital anorectal malformations (CARMs), and whether all patients with CARM need pediatric cardiology screening.

Study design We included 129 patients with CARM born between 2004 and 2013, and referred to University Medical Center Groningen. Recto-perineal and recto-vestibular fistulas were classified as mild CARMs, all others as severe. Significant patent foramen ovale, secundum atrial septal defect, and small ventricular septum defect were classified as minor CHDs, all others as major.

Results Of 129 patients with CARM, 67% had mild CARM, 33% severe CARM, and 17% were additionally diagnosed with CHD. CHDs were distributed equally in patients with mild or severe CARMs. Patients with multiple congenital abnormalities were more frequently diagnosed with CHD (n = 16, 36%) than patients without multiple congenital malformations (n = 5, 9%, $P = .001$). Patients with CARM diagnosed with CHD using pediatric cardiac echo screening were younger than 3 months of age at diagnosis. Earlier general pediatric examinations missed 7 (50%) children with mild and 4 (50%) with severe CHDs.

Conclusions The severity of CARM could predict neither prevalence nor severity of CHD. More than one-half of CHDs were missed during the first physical examination. No new CHDs were found in patients older than 3 months of age at the time CARMs were diagnosed. We recommend screening all patients with CARM younger than 3 months of age for CHD at the time CARM is diagnosed. Preoperative echocardiography should be the rule in children younger than 3 months of age and with multiple congenital anomalies. *(J Pediatr 2016;179:150-3).*

Congenital anorectal malformations (CARMs) occur in approximately one in every 4000 to 5000 live births.[1] These may occur as a single anomaly, as part of the VACTERL association (vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalities) or with congenital syndromes.[2] The presence of CARMs has been associated with congenital heart defects (CHDs).[3-5] The results of previous studies on CHDs in patients with CARMs are ambiguous and report on prevalence varying between 9% and 37%.[2,3] Children with severe forms of CARM are considered to be at greater risk of associated CHDs than children with mild CARMs.[2,4]

Almost two-thirds of CARMs occur in conjunction with other anomalies, such as urinary, genital, and musculoskeletal anomalies, as well as abnormalities and/or defects in the digestive, cardiovascular, and central nervous system.[6,7]

Before any surgery takes place, it is necessary to be aware of the cardiologic condition of the patient to prevent possible complications during anesthesia.[8] Several studies have reported a greater preoperative risk of cardiac arrest and mortality in patients with CHDs undergoing noncardiac surgery.[9-12] CHDs may be overlooked during routine physical examination, although advanced screening techniques with pulse oximetry have improved detection.[13,14]

Our primary goal was to estimate the overall prevalence of CHDs in patients with CARMs, between patients with mild and severe CARMs, and in patients with and without multiple congenital disorders. Finally, we aimed to determine whether preoperative cardiac screening is necessary for all patients with CARM.

Methods

We retrospectively reviewed the medical records of 155 patients with CARMs, who were born between January 2004 and December 2013 and who were referred to the Department of Pediatric Surgery at University Medical Center Groningen, The Netherlands. The study was conducted in compliance with the requirements of our local Medical Ethics Review Board.

We excluded 26 patients; 20 patients had not undergone a full cardiac screening, which included a physical examination and an echocardiogram, by a

CHDs	Congenital heart defects
CARMs	Congenital anorectal malformations

From the [1]Department of Surgery, Division of Pediatric Surgery, University of Groningen, University Medical Center Groningen, Groningen, The Netherlands; [2]Department of Pediatrics, Division of Pediatric Cardiology, University of Groningen, University Medical Center Groningen, Groningen, The Netherlands; [3]Department of Anesthesiology, Division of Pediatric Anesthesiology, University of Groningen, University Medical Center Groningen, Groningen, The Netherlands; and [4]Department of Surgery, Anorectal Physiology Laboratory, University of Groningen, University Medical Center Groningen, Groningen, The Netherlands

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pediatric cardiologist. Six patients were excluded because of missing data. Completion of the medical histories of these 6 patients was impossible for various reasons: 1 patient had re-emigrated, 3 patients were lost because of adoption, and 2 patients died because of severe respiratory insufficiency and/or other physiological failures before screening had taken place. Therefore, our study was based on 129 patients.

Patients with only a rectoperineal or a rectovestibular fistula were classified as having a mild form of CARM, whereas all other patients were categorized as having a severe form.

We also distinguished 2 forms of CHDs: minor CHDs and major CHDs. A significant shunt through a patent open foramen ovale, a secundum atrial septal defect, and/or a small ventricular septal defect were classified as minor CHDs. All other heart defects were categorized as major CHDs.

Patients were defined as having multiple congenital disorders if they have 3 or more congenital disorders, including CARM, or if they had a syndrome that was confirmed by a clinical geneticist.

Statistical Analyses

The data were analyzed with SPSS 22.0 for Windows (IBM SPSS Statistics, IBM Corporation, Armonk, New York). The Fisher exact test was used to compare proportions. CIs were used to estimate the prevalence ratio. The level of statistical significance was set at a probability of <.05.

Results

Out of all 129 patients with CARM, 67% (n = 86) had a mild form of CARM, and 33% patients (n = 43) had a severe form. Fifty-eight percent of the patients were female (n = 75), and 42% were male (n = 54). Among the 129 patients with CARM, 17% (n = 22) were also diagnosed with CHDs (**Table I**).

There was no statistically significant difference in the prevalence of CHD between patients with mild and severe forms of CARM (n = 14, 16% vs n = 8, 19%, $P = .805$). In fact, we found that among patients with mild CARMs, 9% were diagnosed with minor CHDs (n = 8) and 7% with major CHDs (n = 6) ($P = 1.00$). A similar prevalence was found among patients with severe CARMs; 9% had minor CHDs (n = 4) and 9% major CHDs (n = 4) ($P = 1.00$; **Table II**). The prevalence of the CHDs in the different types of CARM are displayed in **Table III** (available at www.jpeds.com).

Out of 102 patients with complete medical data on physical examination, including screening for syndromes, 43% (n = 44) had multiple congenital disorders (**Table I**). We found that patients with multiple congenital disorders had CHDs more often than patients without multiple congenital disorders, 16 (36%) vs 5 (9%), $P = .001$, 95% CI 0.33-0.53, (**Table IV**; available at www.jpeds.com).

The majority of patients with CARM (n = 81, 63%) were diagnosed during the first week after birth. In addition, we found that all patients with CARM (n = 22), who were also diagnosed with CHDs, were younger than 3 months of age at the time of presentation (**Table V**).

Table I. Patient characteristics*

Patients with CARMs N = 129	
Age at CARM diagnosis (mo)	0-73
Female	75 (58%)
Male	54 (42%)
Severe forms of CARM	43 (33%)
Mild forms of CARM	86 (67%)
Multiple congenital disorders (n = 102†)	44 (43%)
Mortality	11 (9%)
Patients with CARM with CHDs	22 (17%)
Patients with CARM without CHDs	107 (83%)

*All patients were born between January 2004 and December 2013 and were referred to the University Medical Center Groningen.
†27 missing values.

Table II. Prevalence of CHD according to CARM severity

Patients with CARMs (N = 129)	Mild CARMs n = 86 (67%)	Severe CARMs n = 43 (33%)	*P* value
CHDs (n = 22, 17%)	14 (16%)	8 (19%)	.805*
Minor CHDs	8 (9%)	4 (9%)	1.000*
Major CHDs	6 (7%)	4 (9%)	1.000*

*Fisher exact test.

Cardiac Screening for Patients with CARM

All patients (N = 129) were given a physical examination and echocardiography by a pediatric cardiologist. Out of the total of 129 patients with CARM, 22 patients were diagnosed with having a CHD before the age of 3 months, and 19 before the age of 7 days (**Tables V** and **VI**). In 90.7% (n = 117) patients with CARM, the routine physical examination had been performed by a general pediatrician prior to the pediatric cardiologist screening. One of the patients with CHD had not received a routine physical examination by a general pediatrician. In 11 (50%) patients, the earlier routine physical examination by a general pediatrician had been performed during the first weeks of life and had not revealed CHDs. Of these 11 patients, 2 (18%) had a major CHD, which was revealed by screening by a pediatric cardiologist. There was no difference between the screening results in mild and severe CARMs.

Discussion

CARMs are known to be associated with CHDs.[1,3-5] Nevertheless, reports on the prevalence of CHD in patients who also

Table V. The number of patients according to age at referral for CARMs and the number of CHDs in these patients

Age at referral	Number of patients	Patients with diagnosed CHDs (n = 22)
0-7 d	n = 81 (63%)	19 (8 major)
<3 mo	n = 30 (23%)	3 (2 major)
>3 mo	n = 18 (14%)	—