Published in final edited form as:

J Perinatol. 2015 October ; 35(10): 842–845. doi:10.1038/jp.2015.74.

Is routine preoperative transthoracic echocardiography necessary in newborns with myelomeningocele?

Margaret M. Vernon, MD¹, Damien Powell, MD^{2,4}, Amy H. Schultz, MD, MSCE¹, Tamara D. Simon, MD, MSPH³, and Dan Doherty, MD, PhD⁴

¹Division of Cardiology, Department of Pediatrics, University of Washington/Seattle Children's Hospital, Seattle, Washington

²Developmental Pediatrics, Madigan Army Medical Center, Tacoma, Washington

³Division of Hospital Medicine, Department of Pediatrics, University of Washington/Seattle Children's Hospital, Seattle, Washington

⁴Division of Developmental Medicine, Department of Pediatrics, University of Washington/Seattle Children's Hospital, Seattle, Washington

Abstract

Objectives—Congenital heart disease is common in newborns with myelomeningocele.

Echocardiography prior to neonatal back closure has been recommended. Its utility in the era of prenatal diagnosis is unclear.

Study Design—We reviewed all newborns with myelomeningocele evaluated by preoperative echocardiography at our institution over 11 years.

Results—76 successive newborns were identified. Ninety-one percent were prenatally diagnosed with myelomeningocele. One percent had critical, 12% critical or noncritical, and 22% critical, noncritical or possible congenital heart disease. The single case of critical congenital heart disease was both prenatally diagnosed and clinically identifiable.

Conclusions—The prevalence of congenital heart disease in newborns with myelomeningocele is increased compared with the general population, however critical disease is uncommon. We propose preoperative echocardiography is unnecessary when the myelomeningocele is prenatally diagnosed, antenatal cardiac screening is complete and normal, and the newborn is clinically well.

Keywords

congenital heart disease; myelomeningocele; echocardiogram; utility

Corresponding Author Contact Information: Margaret M. Vernon, MD, Division of Cardiology, Seattle Children's Hospital, 4800 Sand Point Way NE, RC.2.820, Seattle, Washington 98105, meg.vernon@seattlechildrens.org, Phone: 206-987-7495, Fax: 206-987-3839.

Conflict of Interest: None of the authors have potential financial or personal conflicts of interest to disclose.

The views expressed in this article are those of the authors and do not necessarily reflect the official policy or position of the Department of the Army, Department of the Navy, the Department of the Air Force, Department of Defense, or the US government.

Introduction

Congenital heart disease (CHD) and neural tube defects are two common major malformations linked to folate metabolism¹. Two studies have reported the prevalence of CHD in those with myelomeningocele, a severe defect of neural tube closure resulting in an open spinal canal and dysfunction of the spinal cord and nerves, to be several-fold higher than the general population. In 1999, Ritter *et al* retrospectively studied the perioperative echocardiograms for 105 neonates undergoing myelomeningocele repair in Utah². They reported a prevalence of 4.7% for critical CHD and 37% for all CHD. In 2008, Kocak *et al* similarly reported a prevalence of 5.4% for critical CHD and 40.5% for all CHD in 37 infants with myelomeningocele treated in Turkey.³ Prompt recognition of critical CHD, defined as any cyanotic or ductal dependent lesion, in these newborns is essential as its presence may impact surgical planning and anesthetic management. In response to the first of these publications our institutional standard of care was modified to include a transthoracic echocardiogram prior to myelomeningocele repair. This echocardiogram is usually performed within the first few hours following delivery as back closure is typically performed within 48 hours of birth.

Since the initial work by Ritter *et al.* in 1999, two important changes in practice have occurred, which prompted review of our institutional standard. Today, the vast majority of newborns with myelomeningocele are diagnosed prenatally and, as part of the standard midtrimester anatomic survey, views of the fetal heart are obtained per guidelines published by the American Institute of Ultrasound in Medicine in conjunction with the American College of Radiology and the American College of Obstetricians and Gynecologists.⁴ Secondly, the American Heart Association guidelines on the use of antibiotics for subacute bacterial endocarditis prophylaxis were revised substantially in 2007. Antibiotic prophylaxis is no longer recommended for the majority of congenital lesions, including ventricular septal defects and isolated valve abnormalities.⁵ The objective of this study was to evaluate the need for routine preoperative transthoracic echocardiogram in newborns with myelomeningocele in the era of near-universal prenatal cardiac screening.

Subjects and Methods

Under a research protocol approved by the Seattle Children's Hospital (SCH) Institutional Review Board, we performed a retrospective cohort study of all newborns with myelomeningocele repaired at SCH between January 1, 2000 and December 31, 2011 in whom a preoperative echocardiogram was obtained. SCH is a tertiary care center and the regional neurosurgical referral center for children from five states: Washington, Wyoming, Alaska, Montana and Idaho. Patients were identified for potential inclusion by assignment within the SCH administrative dataset of either an International Classification of Diseases-9th edition- Clinical Modification code 741.xx (all patients with any form of spina bifida) or a Current Procedural Terminology code 63704 or 63706 for repair of myelomeningocele during any inpatient or outpatient visit. Myelomeningocele was defined as a non-skin covered spinal dysraphism in combination with a Chiari II malformation. The list was cross referenced with an internal spina bifida program list to ensure all patients were identified

and included. The composite list was reviewed and all patients with forms of spina bifida other than myelomeningocele excluded.

Patient and maternal demographic information (maternal age, race/ethnicity and gender), gestational age at birth, birth weight, myelomeningocele location, presence of additional malformations not typically associated with myelomeningocele (e.g. equinovarus malformation, spine defects, and Chiari II malformation were not considered additional malformations) and genetic testing results were obtained from the electronic medical record. Preoperative clinical documentation, including the admission note and any consult notes, was reviewed for any clinical findings suggestive of CHD (e.g. cyanosis, murmur, abnormal pulses, persistent tachycardia, tachypnea or respiratory distress and/or oxygen requirement).

Echocardiogram reports were obtained from the electronic medical record, reviewed and findings categorized as critical CHD, noncritical CHD, possible CHD, or normal. Critical CHD was defined as any cyanotic or ductal dependent lesion. Noncritical CHD was defined as any lesion requiring follow-up but unlikely to be hemodynamically significant within the first few days after birth or impact anesthetic management. Examples include atrial and ventricular septal defects and mild valve stenosis. A single cardiologist (AHS) reviewed the archived images (digital or VHS tape if performed prior to 2007) for any report considered indeterminate. An atrial shunt with a tangential trajectory relative to the atrial septum on color flow and no deficiency of septum primum on two dimensional imaging was classified as a patent foramen ovale. An atrial shunt was classified a secundum atrial septal defect if deficiency of septum primum on two dimensional imaging and a perpendicular trajectory of flow relative to the atrial septum on color flow were present. If the shunt remained ambiguous after review, it was classified as possible CHD. Patency of the foramen ovale or ductus arteriosus was considered normal for age. The prevalence of critical CHD, all CHD (critical + noncritical), and all possible CHD (critical + noncritical + possible) was calculated.

Prenatal imaging results were obtained from the newborn or maternal medical record if prenatal counseling occurred as part of the Seattle Children's Prenatal Diagnosis and Treatment Program. In utero cardiac screening was considered complete if the four chamber and both outflow tract views were documented. Study type (e.g. midtrimester anatomic survey or fetal echocardiogram) was not distinguished.

Results

We identified 79 newborns with myelomeningocele who underwent back closure at our institution between January 1, 2000 and December 31, 2011. We excluded three who did not have a transthoracic echocardiogram prior to back closure, leaving 76 (96%) for further investigation (Table 1). The mean $\pm$ SD gestational age and weight at birth were 37 ± 2 weeks and 2.95 ± 0.55 kg, respectively. The study cohort reflected the expected distribution of lesion levels. Ten (13%) of the children, had other congenital anomalies not typically associated with myelomeningocele, and one had a chromosomal abnormality. All back closures were performed within 48 hours of birth, and the average length of stay was 18 days (data not shown).

Seventy-one (93%) newborns appeared clinically well with no documentation of tachycardia, tachypnea, murmur, cyanosis, or hemodynamic instability (Table 1). Four of the five with an abnormal clinical exam were noted to have a murmur on admission to the neonatal intensive care unit with no other signs or symptoms suggestive of congenital heart disease. Two of these four were subsequently diagnosed with non-critical CHD (both ventricular septal defects). The one with a prenatal diagnosis of hypoplastic left heart syndrome was noted to be cyanotic though did not have a murmur on admission. The sensitivity of routine clinical exam to identify CHD in children with myelomeningocele was 33% and specificity was 97% (Table 2).

The preoperative echocardiogram identified or confirmed the presence of CHD in nine newborns (Table 3). One newborn prenatally diagnosed with hypoplastic left heart syndrome was confirmed to have critical CHD (prevalence = 1%) and an additional 8 were diagnosed with noncritical CHD (prevalence of CHD including both critical + noncritical = 12%). Eight newborns had ambiguous atrial level shunts after image review. In total, the prevalence of possible CHD (critical + noncritical + possible) was 22%. Transient ventricular dysfunction was identified in one additional patient. CHD was identified in three of the ten newborns (30%) with an additional congenital malformation, but only 6 of the 66 (9%) without other malformations.

Several consequences of the routine performance of a preoperative echocardiogram were observed. Twelve (16%) preoperative echocardiogram reports identified positioning of the infant to protect the myelomeningocele as a limitation to the study quality or completeness. Nearly all preoperative echocardiograms (72/76 = 95%) identified transitional phenomena (patent ductus arteriosus, patent foramen ovale, or both). Twelve neonates (12/67 = 18%) were serially evaluated by echocardiography despite an absence of CHD. Eleven (92%) of these studies were requested to follow up on transitional phenomena and one subclinical ventricular dysfunction present on the preoperative echocardiogram. All serial evaluations were normal.

The overwhelming majority of newborns admitted with myelomeningocele were diagnosed prenatally (69/76 = 91%) (Table 1). Of the seven newborn diagnoses, three mothers reported no prenatal care or declined both serum and anatomic ultrasound screens, and four reported standard prenatal care including a mid-trimester anatomic survey. Antenatal cardiac imaging results were available in 77% (53/69) of those prenatally diagnosed with myelomeningocele. Two results were considered incomplete, as minimum cardiac screening requirements were not met in the documentation. Critical CHD was diagnosed prenatally in the one fetus with hypoplastic left heart syndrome. *In utero* cardiac screening did not identify many of the atrial or ventricular septal defects (sensitivity 14%) (Table 4).

Discussion

Routine transthoracic echocardiogram screening has been recommended for all newborns with myelomeningocele, however the optimal timing of the echocardiogram has not been specified.^{2,3} At our institution, performance of a preoperative echocardiogram was incorporated into the routine standard of care for those with myelomeningocele. The utility

of this approach has not been evaluated in the current era when the overwhelming majority of newborns with myelomeningocele are diagnosed prenatally (in our cohort over 90%), and screened for CHD *in utero*.⁴ In our retrospective review, the results of antenatal cardiac screening were available in the electronic medical record in 77%. *In utero* cardiac imaging has been shown to reliably detect critical CHD.^{7,8} The sole critical lesion in our cohort was detected prenatally and documentation was readily available to the postnatal care providers in the electronic medical record.

With knowledge of normal *in utero* cardiovascular physiology, our low sensitivity of prenatal cardiac imaging for detecting noncritical CHD is not surprising. An unrestrictive atrial level shunt limits the *in utero* diagnosis of most atrial septal defects and patency of the ductus arteriosus limits the *in utero* diagnosis of many small ventricular septal defects secondary to pressure equilibration between the ventricles.

Routine preoperative transthoracic echocardiogram was highly effective for identifying all forms of CHD, both critical and noncritical in our cohort. Congenital heart disease was identified in 9/76 (12%) and the possibility of CHD was raised in another 8/76 (11%). All total, the possibility of CHD was raised in nearly one quarter of newborns during the preoperative echocardiogram. This multifold higher prevalence of congenital heart disease in newborns with myelomeningocele, in comparison to the general newborn population which has a prevalence of 0.8%, is consistent with, though lower than, previous reports.^{6,2,3} Importantly, critical CHD was uncommon and statistically comparable with prior reports (point estimates range from 1.3 to 5.4%, $p = 0.4$).

Transitional phenomena were identified in nearly all newborns when screening was performed in the preoperative time period. Furthermore, obligate decubitus positioning to protect the back in the preoperative patient resulted in suboptimal imaging in 16% and repeat evaluations were performed in nearly 20% of our cohort to follow up on indeterminate or incomplete findings despite structurally normal hearts. We theorize that many of these serial evaluations could have been avoided if the screening echocardiogram had been delayed until appropriate positioning for a complete study was possible and transitional phenomena have typically resolved.

The newborn examination provides another opportunity to identify CHD. Although the physical examination alone is limited in its ability to identify all forms of congenital heart disease, the physical examination combined with pulse oximetry is highly sensitive, with two studies reporting sensitivities of 82.8% and 93.2%, for detecting critical CHD.^{9,10} In our cohort, the single newborn with critical CHD, in addition to carrying a prenatal diagnosis, was visibly cyanotic and hypoxemia was confirmed with pulse oximetry.

In summary, patients with myelomeningocele appear to have an increased prevalence of CHD; however, critical CHD is rare and can reliably be diagnosed *in utero* when prenatal cardiac screening is performed and complete. Increasingly children's hospital associated multidisciplinary teams provide antenatal consultation when a congenital malformation is identified *in utero* and documentation is readily available to the postnatal care providers in the electronic medical record. We propose that routine preoperative echocardiography is

unnecessary if the myelomeningocele was prenatally diagnosed, documentation of normal *in utero* screening cardiac views, including a four-chamber and both outflow tracts, is available, and the newborn is clinically well including normal pulse oximetry. Conversely, if the myelomeningocele is postnatally diagnosed or documentation of normal *in utero* screening cardiac views is not available, a transthoracic echocardiogram should continue to be performed prior to myelomeningocele repair. Accepting the limitations of antenatal cardiac diagnosis and the increased prevalence of noncritical CHD in those with myelomeningocele, we support the performance of a transthoracic echocardiogram in all newborns with myelomeningocele; however, recommend delaying this examination until after back closure. This approach aims to identify critical lesions likely to impact patient management, while supporting fully integrated, comprehensive, and cost-conscious patient care. Arguably, in those with a normal in utero screen and a normal neonatal physical examination, a postnatal echocardiogram could be excessive, however in our cohort and both previous reports, there appears to be an increased incidence of atrial septal defects in those with myelomeningocele. Atrial septal defects, even when clinically significant in size, are unlikely to be prenatally identified and furthermore are unlikely to be accompanied in the newborn time period by either a murmur or fixed splitting of the second heart sound.¹¹

Looking to the future, the most recent guidelines (2013) endorsed by the American Institute for Ultrasound Medicine, the American College of Obstetrics and Gynecology, and the Society of Maternal Fetal Medicine recommend a fetal echocardiogram be performed in all fetuses with an identified extra-cardiac anomaly.¹² Implementation of this recommendation will lead to a comprehensive *in utero* cardiac evaluation far exceeding the minimum accepted in this study.

Acknowledgements

The authors would like to thank Richard Kronmal, PhD for his statistical expertise, and David Shurtleff, MD and William Walker, MD for their assistance with study design.

Financial Disclosure: TDS is supported by Award K23NS062900 from the National Institute of Neurological Disorders And Stroke, the Child Health Corporation of America via the Pediatric Research in Inpatient Setting Network Executive Council, and Seattle Children's Center for Clinical and Translational Research, and CTSA Grant Number ULI RR025014 from the National Center for Research Resources (NCRR), a component of the National Institutes of Health (NIH). None of the sponsors participated in design and conduct of the study; collection, management, analysis, and interpretation of the data; or preparation, review, or approval of the manuscript. Its contents are solely the responsibility of the authors and do not necessarily represent the official view of the NCRR or NIH.

Abbreviations

CHD	congenital heart disease
NCRR	National Center for Research Resources
NIH	National Institutes of Health
SCH	Seattle Children's Hospital

References

1. Blom HJ, Smulders Y. Overview of homocysteine and folate metabolism. With special references to cardiovascular disease and neural tube defects. *J Inherit Metab Dis*. 2011; 34:75–81. [PubMed: 20814827]
2. Ritter S, Tani LY, Shaddy RE, Minich LL. Are screening echocardiograms warranted for neonates with meningocele? *Arch Pediatr Adolesc Med*. 1999; 153(12):1264–1266. [PubMed: 10591303]
3. Kocak G, Onal C, Kocak A, Karakurt C, Ates O, Cayh S, Yologlu S. Prevalence and outcome of congenital heart disease in patients with neural tube defect. *J Child Neurol*. 2008; 23(5):526–530. [PubMed: 18056696]
4. American Institute of Ultrasound in Medicine. AIUM Practice Guideline for the performance of an antepartum obstetric ultrasound examination. *J Ultrasound Med*. 2003; 22(10):1116–25. [PubMed: 14606571]
5. Wilson W, Taubert KA, Gewitz M, Lockhart PB, Baddour LM, Levison M, et al. Prevention of infective endocarditis: Guidelines from the American Heart Association. *Circulation*. 2007; 116(15):1736–1754. [PubMed: 17446442]
6. Reller MD, Strickland MJ, Riehle-Colarusso T, Mahle WT, Correa A. Prevalence of congenital heart defects in metropolitan Atlanta, 1998-2005. *J Pediatr*. 2008; 153(6):807–813. [PubMed: 18657826]
7. Starikov RS, Bsai FA, Knee AB, Tsirka AE, Paris Y, Markenson GR. Utility of fetal echocardiography after normal cardiac imaging findings on detailed fetal anatomic ultrasonography. *J Ultrasound Med*. 2009; 28:603–608. [PubMed: 19389899]
8. Friedman AM, Phoon CK, Fishman S, Seubert DE, Timor-Tritsch IE, Schwartz N. The utility of fetal echocardiography after an unremarkable anatomy scan. *Obstet Gynecol*. 2011; 118(4):921–927. [PubMed: 21934457]
9. de-Wahl Granelli A, Wennergren M, Sandberg K, Mellander M, Bejrum C, Inganas L, et al. Impact of pulse oximetry screening on the detection of duct dependent congenital heart disease: a Swedish prospective screening study in 39,821 newborns. *BMJ*. 2009; 338a:3037.
10. Zhao Q, Ma XJ, Xiao-ling G, Liu F, Yan W, Wu L, et al. Pulse oximetry with clinical assessment to screen for congenital heart disease in neonates in China: a prospective study. *Lancet*. 2014; 384:747–54. [PubMed: 24768155]
11. Porter, CJ.; Feldt, RH.; Edwards, WD.; Seward, JB.; Schaff. Atrial septal defects. In: Allen, HD.; Clark, EB.; Gutgesell, HP.; Driscoll, DJ., editors. *Moss and Adams' Heart Disease in Infants, Children, and Adolescents: Including the Fetus and Young Adult*. 7th. Lippincott Williams & Wilkins; Philadelphia, PA: 2008. p. 632-645.
12. American Institute of Ultrasound in Medicine. AIUM practice guideline for the performance of fetal echocardiography. *J Ultrasound Med*. 2013; 32:1067–1082. [PubMed: 23716531]

Table 1

Patient characteristics

Characteristic	^I n	%
Gestational age: mean $\pm$ SD in weeks (range)	37 $\pm$ 2 (32 – 40)	
Birth weight: mean $\pm$ SD in kg (range)	2.95 $\pm$ 0.55 (1.3 – 4.4)	
Sex		
Male	34	45
Female	42	55
Myelomeningocele location		
Thoracic	2	3
Thoracolumbar	6	8
Lumbar	28	37
Lumbosacral	30	39
Sacral	10	13
Abnormal genetic testing	1	
Additional birth defects	10	13
Low set ears	1	
Preauricular skin tags	1	
Limb abnormalities (nonequinovarus)	1	
Genitourinary	3	
Gastrointestinal	1	
Diaphragmatic hernia	1	
Polydactyly	1	
Hemivertebrae	1	
Cardiovascular exam documentation		
Normal	71	93
Abnormal ²	5	7
Prenatal diagnosis of myelomeningocele		
Yes	69	91
No	7	9

¹Data expressed in number of patients unless otherwise indicated

²Included documentation of tachycardia, tachypnea, murmur, cyanosis, or hemodynamic instability

Table 2

Sensitivity and specificity of routine clinical evaluation for CHD

	CHD (critical + noncritical) (n = 9)	No CHD ^I (n = 67)
Abnormal clinical evaluation	3	2
Normal clinical evaluation	6	65

Sensitivity of routine clinical evaluation = $3/9 = 33\%$ Specificity of routine clinical evaluation = $65/67 = 97\%$ ^IPatients with possible CHD, ie) ambiguous atrial level shunts, are included in this category

Table 3

Prevalence of CHD on preoperative echocardiograms (n=76)

Echocardiographic diagnosis	Frequency	%
Critical CHD	1/76	1
Noncritical CHD	8/76	11
Ventricular septal defect ¹	6	
Atrial septal defect	1	
Valvar abnormality	1	
Double aortic arch ¹	1	
All CHD (critical + noncritical)	9/76	12
Possible CHD ²	8/76	11
All possible CHD	17/76	22

¹ 1 child had a ventricular septal defect and double aortic arch and is counted once in the total with CHD but twice in lesion specific tally

² Ambiguous atrial level shunt

Table 4

Sensitivity and specificity of antenatal cardiac imaging for detecting CHD (n=51)

	CHD (critical + noncritical)	No CHD ^I
Abnormal cardiac screen	1	0
Normal cardiac screen	6	44

Sensitivity of detailed fetal anatomic ultrasound = $1/7 = 14\%$ Specificity of detailed fetal anatomic ultrasound = $44/44 = 100\%$ ^IPatients with possible CHD, ie) ambiguous atrial level shunts, are included in this category