

The Utility of Screening Echocardiograms in Congenital Chest Wall Deformities

Keywords: Congenital chest wall deformities; connective tissue disease; echocardiograms; screening tests; pectus excavatum; pectus carinatum

Abstract

Objective: Congenital chest wall deformities (CCWD) are bony or cartilaginous defects of the chest. These deformities result in both an abnormal appearance and, in many cases, cardiopulmonary compromise. Evidence is growing regarding the cardiovascular implications of CCWD. Screening echocardiograms (ECHOs) are increasingly utilized for patients with chest wall deformities to assess cardiac function. We sought to describe the impact of screening ECHOs on patients with CCWD.

Methods: This is a retrospective study. Using ICD10 diagnosis codes, a cohort was generated. Manual chart review performed for demographic data, CCWD diagnosis, and ECHO reports. Additional chart review was performed to see if patients were seen by Pediatric Cardiology, and what the documented plan was including potential intervention. Results: We found 494 patients that fit inclusion criteria. Patients with CCWD most often had pectus excavatum or pectus carinatum. The most common ECHO findings included mitral valve prolapse, patent foramen ovale, and aortic root dilation. Most patients did not require cardiology follow up (86%), with even less requiring cardiologic interventions (1%). We found 10 patients with connective tissue disease.

Conclusions: Patients with CCWD benefit from screening ECHOs in order to find cardiac anomalies that could impact pre, intra, and post-operative management. Findings could also initiate the workup for CTD and Cardiology involvement, further improving patient care.

Glossary of Abbreviations

1. Congenital chest wall deformities - CCWD
2. Pectus Excavatum - PE
3. Pectus Carinatum - PC
4. Pectus Arcuatum - PA
5. Poland Syndrome - PS
6. Connective Tissue Disorder - CTD
7. Echocardiogram - ECHO
8. Left ventricle ejection fraction - LVEF
9. Mitral valve prolapse - MVP
10. Patent foramen ovale - PFO
11. Aortic root dilation - ARD
12. Marfan's Syndrome - MFS
13. Ehlers-Danlos Syndrome - EDS
14. Loeys-Dietz Syndrome - LDS
15. Mixed Connective Tissue Disease - MCTD



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16. Anterior-to-Posterior - AP
17. Right ventricle - RV
18. Ventricular premature complexes - VPC

Introduction

Congenital chest wall deformities (CCWD) are bony or cartilaginous defects of the chest. This includes pectus excavatum (PE), pectus carinatum (PC), pectus arcuatum (PA), and Poland syndrome (PS). These occur in 1 in 300 to 1000 live births [1]. PE and PC are the most common, and present with a depression and protrusion of the chest wall, respectively. These deformities result in both an abnormal appearance and, in many cases, cardiopulmonary compromise. Multicenter studies have shown that patients with these deformities have negative body image that improves following surgical correction [2]. There is growing and substantial evidence regarding the cardiovascular implications of CCWD. The severity of cardiovascular symptoms ranges from mild, including palpitations or chest pain, to severe, such as life-threatening arrhythmias [2]. Most commonly patients report exercise intolerance and dyspnea.

While CCWD are an isolated entity, they can be indicative of underlying connective tissue disorders (CTD). The most associated CTD with CCWD are Noonan syndrome and Marfan syndrome. Both syndromes are inherited in an autosomal dominant fashion and can have various cardiac defects including valvular abnormalities, cardiomyopathies, and aortic dilation or dissection [3]. While certainly not all CCWD have a direct link to CTD, this close association heightens suspicion for potential underlying cardiac defects.

Screening echocardiograms (ECHOs) are increasingly utilized for patients with chest wall deformities to assess cardiac compression, function, and screen for underlying cardiac anomalies. Several studies have evaluated the implications of pectus deformities on the heart such as mitral valve prolapse, reduced right ventricle filling, and reduced cardiac output [4]. These are most significant in PE patients where the heart can be displaced or depressed secondary to the indentation of the chest wall. These anomalies could contribute to

the most reported symptoms of chest pain, palpitation, and exercise intolerance by pectus patients. Given the association of CCWD and CTD, screening ECHOs also help identify cardiac or aortic anomalies that need to be addressed before pectus correction.

This study aims to further our understanding of pectus deformities impact on the cardiovascular system by analyzing echocardiographic data. We additionally sought to elucidate the impact of screening ECHOs in those with positive findings. This was achieved by examining ECHO reports for positive findings, and if these findings warranted cardiology visits, additional follow up. Findings from this study could shape cardiovascular screening, cardiology referral practices, and monitoring protocols in this patient population.

Methods

Cohort Generation

This was a retrospective study. All patient gathering was performed after IRB approval (IRB# 17618). Informed consent was waived given the retrospective nature of the study and all patient information being de-identified after collection. Inclusions criteria included all patients seen in the Oklahoma Children’s Hospital Congenital Chest Wall Clinic between July 1st, 2020 through July 1st, 2024 between the ages of 0 to 17. All patients were seen and managed by a single provider during this time. Patients were selected based on ICD 10 code diagnosis. The codes used were: Q67.6, Q67.7, Q67.8, Q76.6, Q76.7, Q76.8, Q76.9, and M95.4. Exclusion criteria included those who had seen the provider for reasons other than our inclusion criteria, those older than 17, and those without ECHO reports.

This list of patients was then uploaded to REDCap. We performed manual chart review. Demographic data gathered included age at initial clinic visit and gender. CCWD diagnosis was confirmed, and ECHO reports reviewed for structural abnormalities and ejection fraction. Further chart review was done for if the patient was seen by Pediatric Cardiology, and what the documented plan was at their initial visit. Additionally, it was noted if the patient has had any Cardiology based intervention such as started medications, catheterizations, or pacemaker placements. Finally, patient charts were examined to see if the patient had a CTD diagnosis.

Statistical Analysis

Descriptive statistics were used to summarize the data. Bivariate testing of associations was performed using chi-square tests. T-tests were used to evaluate differences in continuous variables. All data management and analyses were performed using SAS v9.4 (SAS Institute, Cary, NC). Significance was set at a value of p=0.05.

Results

Cohort

A total of 676 patients were found (Figure 1). Of these, 182 were excluded for incomplete ECHO data, or ECHOs being reported as shortened interpretations in clinic notes instead of formal reports by faculty when full reports were unavailable (n= 494), (Table 1). The latter usually had pertinent findings but usually did not include LVEF. The cohort was vastly male (83.8%) and the median age was 13 (mean 12.3, mode 13, std. dev 3.78). Over half of the patients were seen for PE, followed closely by PC.

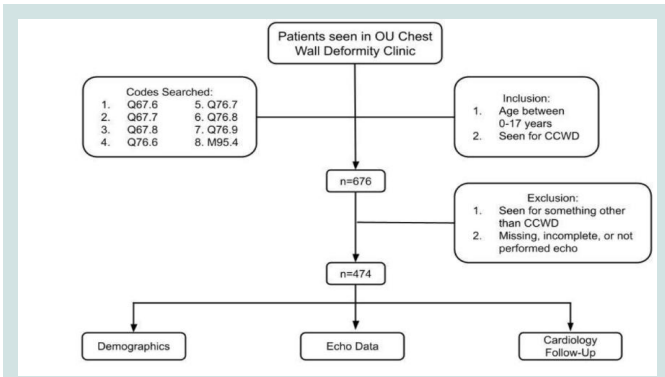


Figure 1: Cohort Generation

Table 1: Demographic Data

Demographic Data			
Gender		n	Percent (%)
	Male	414	83.8%
	Female	80	16.2%
Age			
	Median	13	
	Mean	12.27	
	Mode	13	
	Std. Dev.	3.78	
CCWD		n	Percent (%)
	PE	260	52.6%
	PC	218	44.1%
	PA	5	1.01%
	Other	5	1.01%
	Mixed pectus	4	0.81%
	Acquired	2	0.41%

Echocardiogram Findings and Cardiology Evaluation

The median LVEF was 64% (mean 64.49%, std dev 6.09; (Table 2). There were 53 positive screening ECHOs (10.7%), with 75 total findings (as some patients had multiple findings). In PE and PC, the most common anomalies included: Mitral valve prolapse (MVP), Patent foramen ovale (PFO), aortic root dilation (ARD). Abnormalities were most often seen in PE (PE 13%, PC 7%; p=0.032). All cases were newly diagnosed and were not a pre-existing condition for these patients.

Most patients were not seen by a Cardiologist based on their ECHO findings (n=425, 86.0%; (Table 3). For those seen by Cardiology, common plans included repeat followups up at various intervals (1, 2 or 3 years usually), repeating an ECHO with follow up, outpatient monitors for arrhythmias, and genetics referrals for CTD workup. Of the few seen by Cardiology, even less required cardiologic interventions (n=5, 1%). Interventions included heart catheterization, starting medications (atenolol and losartan), two radiofrequency ablations, and one dual chamber transvenous pacemaker placement.

Connective Tissue Disease Prevalence

Ten patients were found to have CTD (2.02%) (Table 4). We found four different CTDs in our patient population. These include Marfan Syndrome (MFS; n=4), Ehlers-Danlos Syndrome (EDS; n=3), Loeys-Dietz Syndrome (LDS; n=2), and Mixed CTD (MCTD; n=1).

Table 2: Echocardiogram Data

Echocardiogram Data			
Ejection Fraction			
	Median	64	
	Average	64.49	
	Std Dev	6.09	
Abnormal Findings		n	Percent (%)
	Mitral valve prolapses	16	3.24%
	Patent foramen ovale	15	3.04%
	Aortic root dilation	12	2.43%
	Aortic insufficiency	7	1.42%
	Ventricular septal defect	5	1.01%
	Aortic valve anomaly	3	0.61%
	Left ventricular hypertrophy	3	0.61%
	Atrial dilation	3	0.61%
	Ventricular dilation	3	0.61%
	Coronary artery fistula	2	0.41%
	Patent ductus arteriosum	1	0.20%
	Right atrial/ventricular enlargement	1	0.20%
	Restrictive cardiomyopathy	1	0.20%
	Pulmonary artery dilation	1	0.20%
	Atrial septal defect	1	0.20%
	Ascending aorta dilation	1	0.20%
	Sum	75	15.20%

Table 3: Cardiology Information

Cardiology Information			
Had Cardiology follow-up?		n	Percent (%)
	No	425	86.0%
	Yes	69	14.0%
Required Cardiology intervention?		n	Percent (%)
	No	489	99.0%
	Yes	5	1.01%

Patients with CTD had either PE (n=5; 50%), PC (n=4; 40%) or PA (n=1; 10%).

Discussion

CCWD are a diverse family of pathologies. This group of diseases are often associated with underlying cardiac abnormalities, and have a predisposition to CTD such as MFS or EDS. Through our study, we better describe the relationship between CCWD and associated findings on screening ECHOs, further validating the need for preoperative screening.

A recent study by Pitt *et. al.* examined 1510 patients with CCWD. They found that 80% of their patients were male, which is similar to our findings. PE was the most common CCWD between our two studies at ~50%, followed closely by PC at ~45% and then all others [5]. Previous research by our group focused on PC. In that study there were 155 patients with PC, and 10% were found to have a cardiac anomaly. These were most usually MVP and aortic root dilation [6]. Of the 228 patients with PC, 16 had a positive ECHO finding. The most common were PFO, ARD, and MVP. Other than a slightly lower incidence of positive findings, this is in line with the previously mentioned study.

PE was significantly more often associated with positive ECHO findings compared to PC. There were not enough instances of all CCWD to make any meaningful conclusions. A recent systematic review by Sonaglioni *et. al.* found that children with PE had a three-fold higher chance of having MVP compared to adults. This is thought to be due to the smaller thoracic volume of children, resulting in more anterior-to-posterior (AP) compression of the right ventricle (RV) and great distortion of the mitral valve annulus [7].

While most of these findings may be mild, they have the potential to significantly impact the perioperative period. The altered sternal body of PE can cause RV compression and decreased stroke volumes due to insufficient filling [8-10]. MVP is also associated with increased incidences of arrhythmias such as ventricular premature complexes (VPCs, [11]).

CCWDs’ association with CTDs puts patients at a higher risk for cardiac anomalies. PE has been associated with 20 unique genetic disorders, of which 5 of primary CTD [12]. In a study by Rhee *et. al.*, patients with PE had increased rates of ARD. Further genetic evaluation of those with ARD found that 2 of 5 patients were newly diagnosed with MFD [13]. Depending on the degree of ARD, this can increase operative and anesthetic risk. A study by Elsäcker *et. al.*, that ARD become more pronounced at 8 years old. Those who underwent intervention for ARD usually happened around 16 years of age [14]. Finding these patients early could allow for earlier medical or surgical intervention, or closer surveillance of aortic root diameter.

Our study has its limitations. Our patient cohort was generated from a single center and only pediatric patients. This can limit generalizability to other locations, as well as to adult patients. While every patient in our study had an ECHO ordered, not all patients had an ECHO on file. This could be due to a recent transition in our electronic medical record (EMR) system. Other reasons for this included that a screening ECHO was not indicated at the time due to patient age, or that it was obtained at an outside facility with results not uploaded to the EMR.

Future directions would be continued analysis of patients. This would increase the power of the study and help draw more meaningful conclusions. We could also further bolster our study by partnering with others for a multi-institutional study. Doing so would again increase power and would also make our results more generalizable. Finally, another common alteration of CCWD includes arrhythmias. During chart review, we did note that some patients who saw cardiology were noted to have positive EKG findings, but these were noted in an informal way. A formal evaluation of EKGs as an additional screening tool in this patient population is warranted.

Another potential area of interest would be cost-benefit analysis. ECHOs can cost anywhere from \$204 to \$2,588 depending on the patient’s insurance [15]. Given those with CCWD have a higher risk of CTD and CTD patients are at the highest risk during operative intervention, this population would be of particular interest.

Conclusion

Based on this study, we will continue to pursue preoperative screening ECHOs in all patients with CCWD. While the incidence of positive findings is low, the impact of the findings can greatly impact

Table 4: Connective Tissue Disease Data

Connective Tissue Disease Data										
CTD diagnosis?		n	Percent (%)		Diagnosis + CCWD Type		PE	PC	PA	Total
Yes		10	2.02%		Marfan's Syndrome		2	2	0	4
No		484	98.0%		Loeys-Dietz		2	0	0	2
Diagnosis		n	Percent (%)		Ehlers-Danlos		0	2	1	3
Marfan Syndrome		4	40%		Mixed CTD		1	0	0	1
Ehlers-Danlos		3	30%		Total		5	4	1	10
Loeys-Dietz		2	20%							

pre, intra, and post-operative management of this patient population. Positive findings can prompt evaluation by cardiology, allowing for preoperative risk stratification and long-term follow-up if indicated. Screening ECHOs can also help start the process of genetic screening for CTDs, further improving patient care.

References

1. Kloth K, Klohs S, Bhullar J, Boettcher M, Hempel M, et al. (2022) The Epidemiology behind Pectus Excavatum: Clinical Study and Review of the Literature. *Eur J Pediatr Surg* 32: 316-320.

2. Dean C, Etienne D, Hindson D, Matusz P, Shane Tubbs R, et al. (2012) Pectus Excavatum (funnel chest): A Historical and Current Prospective. *Surgical and Radiologic Anatomy* 34: 573-579.

3. Kotzot D, Schwabegger AH (2009) Etiology of Chest Wall Deformities – A Genetic Review for the Treating Physician. *Journal of Pediatric Surgery* 44: 2004-2011.

4. Maagaard M, Heiberg J (2016) Improved cardiac function and exercise capacity following correction of pectus excavatum: a review of current literature. *Ann Cardiothorac Surg* 5: 485-492.

5. Pitt JB, Zeineddin S, Carter M, Perez A, Sands L, et al. (2024) Demographics of Anterior Chest Wall Deformity Patients: A Tertiary Children's Hospital Experience. *J Surg Res* 293: 451-457.

6. Port E, Hunter CJ, Buonpane C, Vacek J, Sands L, et al. (2020) Echocardiography Reveals Heart Abnormalities in Pediatric Pectus Carinatum. *Journal of Surgical Research* 256: 364-367.

7. Sonaglioni A, Bruno A, Polymeropoulos A, Nicolosi GL, Lomarbo M, et al. (2024) Prevalence of Mitral Valve Prolapse Among Individuals with Pectus Excavatum: A Systematic Review and Meta-Analysis. *Diagnostics* 14: 2488.

8. Mao YZ, Tang S, Li S (2017) Comparison of the Nuss versus Ravitch Procedure for Pectus Excavatum Repair: An Updated Meta-Analysis. *Journal of Pediatric Surgery* 52: 1545-1552.

9. Obermeyer RJ, Cohen NS, Jaroszewski DE (2018) The Physiologic Impact of Pectus Excavatum Repair. *Seminars in Pediatric Surgery* 27: 127-132.

10. Ganescu O, LaRusso K, St-Louis E, Saint-Martin C, Cavalle-Garrido T, et al. (2022) The Utility of Echocardiography and Pulmonary Function Testing in the Preoperative Evaluation of Pectus Excavatum. *Journal of Pediatric Surgery* 57: 1561-1566.

11. Kavey R-EW, Blackman MS, Sondheimer HM, Byrum CJ (1984) Ventricular Arrhythmias and Mital Valve Prolapse in Childhood. *The Journal of Pediatrics* 105: 885-890.

12. Billar RJ, Manoubi W, Kant SG, Wijnen RMH, Demirdas S, et al. (2021) Association between Pectus Excavatum and Congenital Genetic Disorders: A Systematic Review and Practical Guide for the Treating Physician. *Journal of Pediatric Surgery* 56: 2239-2252.

13. Rhee D, Solowiejczyk D, Altmann K, Prakash A, Gersony W, et al. (2008) Incidence of Aortic Root Dilation in Pectus Excavatum and Its Association with Marfan Syndrome. *Archives of Pediatrics & Adolescent Medicine* 162: 882-885.

14. Van Elsacker E, Vink A, Menke L, Pals G, Bokenkamp R, et al. (2022) Growth of the Aortic Root in Children and Young Adults with Marfan Syndrome. *Open Heart* 9: e002097.

15. Oseran A, Ati S, Feldman WB, Gondi S, Yeh R, et al. (2022) Assessment of Prices for Cardiovascular Tests and Procedures at Top-Ranked US Hospitals. *JAMA Internal Medicine* 182: 996-999.nces: