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REVIEW ARTICLE

Can Functional Echocardiography be Useful in the Diagnosis of Congenital Heart Disease? Review of Two Cases of Transposition of Great Arteries

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ABSTRACT:
Introduction: Congenital heart diseases represent the most frequent form of malformation and can be diagnosed by fetal echocardiography, neonatal screening or clinical signs. Whenever this is suspected, a pediatric cardiologist should be contacted, and comprehensive echocardiography performed. But the availability of a specialist for immediate implementation is not the reality of many services. As functional echocardiography is often carried out in neonatal units because of its utility in several clinical situations, we consider its usefulness in diagnosing congenital heart diseases. Case reports: we report two cases of transposition of the great arteries diagnosed by functional echocardiography. Case 1 was a premature neonate transferred to the neonatal unit with a suspected diagnosis of respiratory distress syndrome and sepsis without improvement with initial treatment. Functional echocardiography on admission allowed the diagnosis of transposition of the great arteries. Case 2 was a full-term neonate with early cyanosis, with functional echocardiography suggesting transposition of the great arteries and the images used for discussion with a specialist from a reference center, speeding up the transfer. Discussion: early recognition of critical heart disease is essential to improve prognosis. Diagnosis of these diseases can be performed by fetal echocardiography, neonatal screening and clinical signs. When congenital heart structural defect is likely, pediatric cardiologist should be consulted and comprehensive echocardiography promptly performed. Functional echocardiography has been performed in neonatal units with the aim of analyzing the patent ductus arteriosus and ventricular function, diagnosing cardiac tamponade and neonatal persistent pulmonary hypertension. As not all services have a pediatric cardiologist readily available, we considered whether functional echocardiography could help in the diagnostic conclusion through systematic analysis of cardiac images. **Conclusion:** we reinforce that comprehensive echocardiography performed by a specialist is the first option or performed as soon as possible when congenital heart disease is the main hypothesis.

Keywords: neonatology, congenital heart disease, functional echocardiography.

1) Introduction:

Cardiovascular disorders are common in the neonatal period, usually secondary to congenital heart defects, arrhythmias, myocardial dysfunction or complications of other neonatal disorders. The evaluation is based on clinical data and vital signs, but this is not always sufficient to clarify the diagnosis. Use of Point-of-Care ultrasound may help with additional information. With respect to the target neonatal echocardiogram (TnECHO), there are clear limitations when there is a potential for congenital heart disease. Congenital heart disease should be considered when fetal echocardiography shows an abnormality, when a neonatal screening test is positive or equivocal, or when clinical evidence suggests a structural defect. In the neonatal period, critical cardiopathies may manifest with early cyanosis or signs of systemic malperfusion. [1-4] Thus, European Echocardiography Societies suggest that imaging specialist should perform an examination as early as possible. [5]

In recent decades, point-of-care ultrasound (POCUS) has developed and become a common practice in intensive care and emergency units.[4] Its principle complements the clinical examination and answers simple questions or assists in performing invasive procedures. In many services of developing countries, imaging specialists are not always readily available. In these cases, cardiac ultrasound is performed by an imaging non specialist before the specialist.

Several congenital heart diseases can be hard to diagnose even for the specialist, such as anomalous drainage of the pulmonary veins and coarctation of the aorta. [1] On the other hand, a professional with advanced training in functional echocardiography rarely lacks signs that can identify structural cardiac pathology, particularly taking into account the clinical data.[2]

2) Case Reports:

Review of two cases of congenital heart disease diagnosed by functional echocardiography and review of the literature.

Case 1: neonate transferred from a city located 300 km away, premature (32 weeks of gestational age), weighing 1,745g, mother being treated for a urinary tract infection and showing signs of chorioamniotitis, Apgar 6/7, progressing to tachydyspnea. Continuous positive airway pressure (CPAP) was installed and subsequently intubated and administered surfactant, mechanical ventilation with high parameters and antibiotic therapy. Having little response, the surfactant was repeated and transferred at 36 hours of life. On admission to the intensive care unit, he was sedated, 80% saturation despite adequate chest expansion and 100% inspired oxygen fraction. Functional echocardiography was performed, which demonstrated the absence of crossing of the great vessels in the subcostal window and the visualization of a bifurcation vessel in the apical window. Prostaglandin started immediately, managed to evolve with a reduction in mechanical ventilation, requested a comprehensive echocardiogram by a specialist who confirmed the hypothesis of transposition of the great arteries. Stabilized and transferred to cardiovascular surgery service.

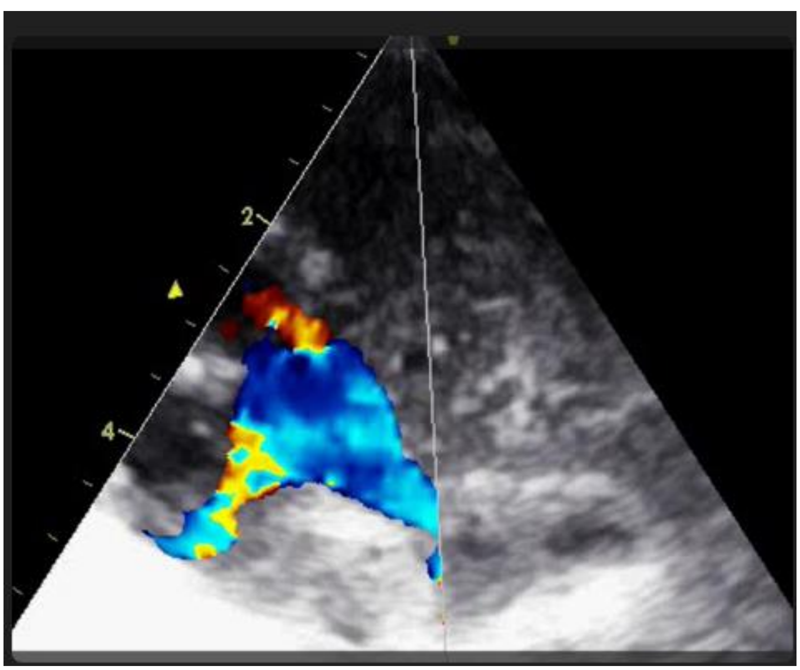

Figure 1: left ventricular outflow tract, showing a bifurcating vessel (pulmonary artery).

Case 2: neonate at 39 weeks of gestational age, elective cesarean delivery, weight 3,660g, Apgar 8/8 evolved with tachypnea and central cyanosis on the first day of life, maintaining saturation below 80% even with a 60% oxygen helmet. He was taken to the intensive care unit and intubated, coupled to mechanical ventilation. When performing functional echocardiography, a bifurcation vessel was observed in the left ventricular outflow tract, compatible with the hypothesis of transposition of the great arteries. Started prostaglandin. Image sent to a child cardiologist in charge of a heart surgery reference service, who accepted transfer even without echocardiographic evaluation by the specialist.

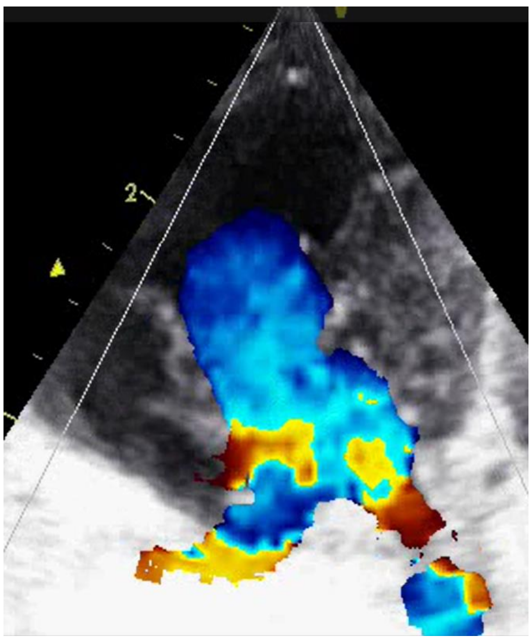

Figure 2: Vessel that bifurcates in the left ventricular outflow tract.

3) Discussion:

Congenital heart disease (CHD) is the most common type of congenital anomaly, with an overall prevalence of 1%.[6] Critical CHD, defined as lesions that require surgery or catheter intervention in the first year of life, account for approximately 25% of defects cardiac structures. Many newborns with critical CHD are diagnosed prenatally or are identified shortly after birth (due to signs/symptoms or positive pulse oximetry screening). In infants with critical heart injuries, the risk of morbidity and mortality increases when there is a delay in diagnosis and timely referral to a tertiary center specializing in the care of these patients. [6]

Congenital heart diseases present with signs and symptoms common to several other common diseases of the neonatal period, in addition to the very process of transition from fetal to neonatal circulation being able to confuse the reasoning of the professionals responsible for the assistance. It is therefore essential that the pediatrician has a good knowledge of the evocative manifestations of heart disease.

Among patients with a high index of suspicion for the anatomical heart disease, there is no discussion that echocardiography should always be performed in consultation with the pediatric cardiology team. In critically ill patients in whom the suspicion of complex CHD is not high, neonatal functional echocardiography can be used to help stabilize the hemodynamic state.[7] Functional echocardiography is part of point-of-care ultrasound, when the imaging nonspecialist performs the insonation to complement the clinical examination, aiding diagnoses and/or facilitating medical procedures.

Functional echocardiography is used in neonatal units to evaluate the patent ductus arteriosus, evaluate cardiac hemodynamics, analyze neonatal persistent pulmonary hypertension, investigate cardiac tamponade and locate catheter tips. Recently, a study demonstrated that after training in functional echocardiography, neonatologists achieved good accuracy in functional assessments when compared to specialists.[7] A study carried out by Bischoff and collaborators demonstrated high diagnostic agreement between neonatologists trained in functional echocardiography and pediatric cardiologists with experience in imaging. This study highlights that intensivist, with comprehensive and rigorous training in targeted neonatal echocardiographic methods, can provide timely and clinically effective neonatal cardiovascular care without major safety concerns when following guidelines for evaluating infants with a low suspicion of CHD. The authors highlighted the importance of a collaborative and integrated approach between the neonatal intensive care unit and the pediatric echocardiography laboratory and the need to develop quality assurance standards.[8]

Thus, functional echocardiography should not be performed with the aim of diagnosing congenital heart diseases. Whenever there is this possibility, a specialist should be requested for evaluation and comprehensive echocardiography. However, this professional will not always be readily available. Other times, the initial hypothesis is other pulmonary diseases or related to the transition of the fetal circulation. In these cases, functional echocardiography would be indicated and may help in the diagnostic definition; but the professional who performs the exam needs to be aware of alterations that indicate a structural defect, in addition to considering the limitations of the exam. We describe two cases of critical heart disease diagnosed by a neonatologist trained in point-of-care ultrasound.

The first case is related to a premature neonate with a risk factor for sepsis and poor response to the use of pulmonary surfactant. The team that conducted the case in the city of origin thought about respiratory distress syndrome and early sepsis, having established a treatment based on these hypotheses. The reason for the transfer was the poor response and the need for high ventilatory parameters, maintaining low saturation. Neonatal persistent pulmonary hypertension could be the cause of inadequate response, which could be confirmed by echocardiography with evaluation of right ventricular dynamics and estimated measurement of pulmonary artery pressure in the presence of tricuspid regurgitation. Upon admission to the ICU, functional echocardiography was performed immediately and interpreted by the examiner as transposition of great arteries.

In the second case, there is a typical description of early-onset congenital heart disease with cyanosis. In this case, obstruction of the right ventricular outflow tract with right-to-left intracardiac shunt and transposition of the great arteries are the initial hypotheses, with the prostaglandin indicated for opening the ductus arteriosus and immediate request for echocardiography by a specialist. In this case, obtaining images by functional echocardiography made it possible to send the images to the specialist and discuss the case by telephone, favoring agility in the transfer of the newborn.

When functional echocardiography is performed before the analysis by the child cardiologist and comprehensive echocardiography, it is necessary to perform a systematic examination, using all the windows that allow identifying suspicious alterations. Pediatric cardiologists recommend sequential segmental analysis as a diagnostic basis of congenital heart diseases. This analysis assumes that all hearts, normal or abnormal, are made up of atria, ventricles and arteries. The way in which these structures are related must be analyzed. One of the main characteristics of many malformed hearts is that these structures are not always in the usual place.[9,10] Therefore, analyze whether there is atrioventricular and ventriculoarterial concordance. The first step is to analyze whether the *situs* is *solitus* and, if so, the aorta is positioned on the left and the vena cava on the right. When analyzing the subcostal window, start with the transducer at 90^0 in relation to the patient's skin with the marker oriented to the left, identify the aorta and vena cava and confirm that it is *situs solitus* (figure 3).

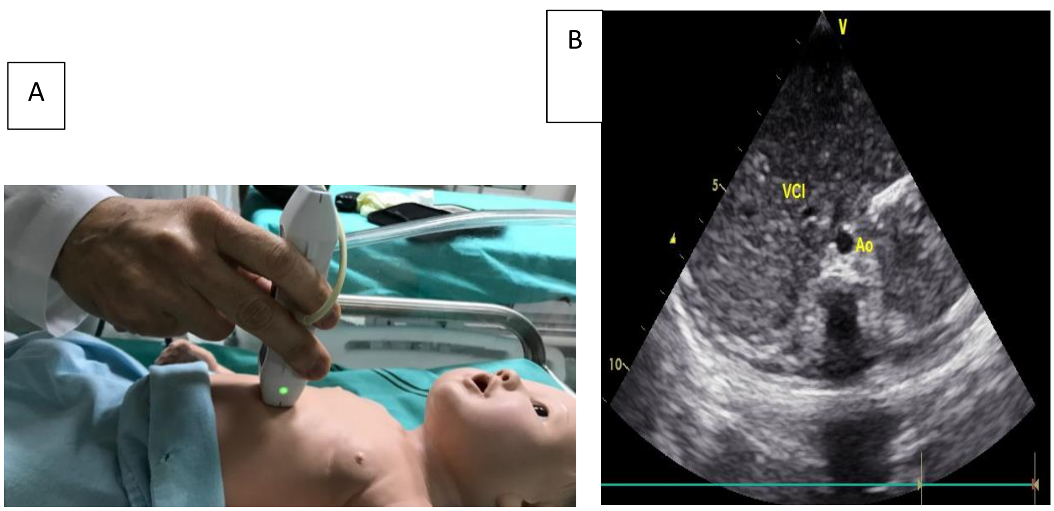

Figure 3: Subcostal window. A) Transducer positioning at 90° and marker facing left. B) Image of the aorta (Ao) and inferior vena cava (IVC).

To assess the atria, the probe is positioned below the xiphoid process in an axial fashion with the marker pointing to the left. The beam is then angled towards the anterior chest wall until the atria come into view. This is the best window for analyzing the interatrial septum. Analyze with color flow and observe the flow direction. Further anterior angulation of the beam will reveal the outflow tract of the left (LVOT), and then the right (RVOT) ventricle. That is, by continuing to move the bundle anteriorly, the aorta disappears, and the pulmonary artery emerges from the right ventricle (crossing of the vessels, figure 4).[9,10] In the transposition of the great arteries, when placing the transducer anteriorly, the crossing is not seen, because the two vessels are in parallel. [9]

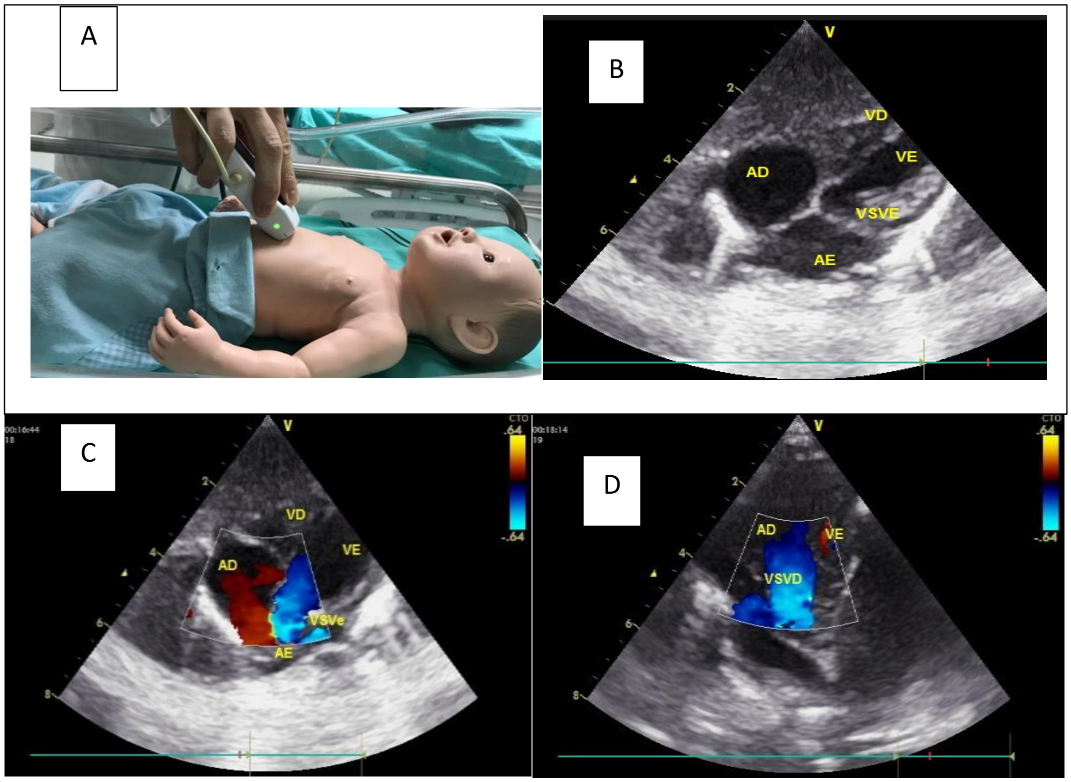

Figure 4: Subcostal window. A) Positioning of the transducer. B) Visualization of the cardiac chambers and analysis of the interatrial septum. C) With the transducer anterior, visualization of the left ventricular outflow tract. D) By continuing to move the transducer anteriorly, view the right ventricular outflow tract and observe the crossing of the vessels. AD: right atrium, AE: left atrium, RV: right ventricle, LV: left ventricle, RVOT: right ventricular outflow tract, SVVe: left ventricular outflow tract.

When analyzing the apical 4-chamber window, identify whether there is atrioventricular concordance. The right ventricle has an atrioventricular valve more "inside" the ventricle and has a moderator band. Use color flow to identify whether there is a shunt between ventricular chambers and its direction. Also, observe the flow through the atrioventricular valves. Search for valve regurgitation. Attention at this point because tricuspid regurgitation may be present in pulmonary hypertension, but it will also be present in obstructions in the right ventricular outflow tract. Interventricular communication is identified in all echocardiographic planes. To bring the left ventricular outflow tract and aortic valve into view, the probe is rotated in a clockwise motion and angled slightly anteriorly. Further anterior angulation will bring the right ventricular outflow tract and the pulmonary valve into view. [9-11]

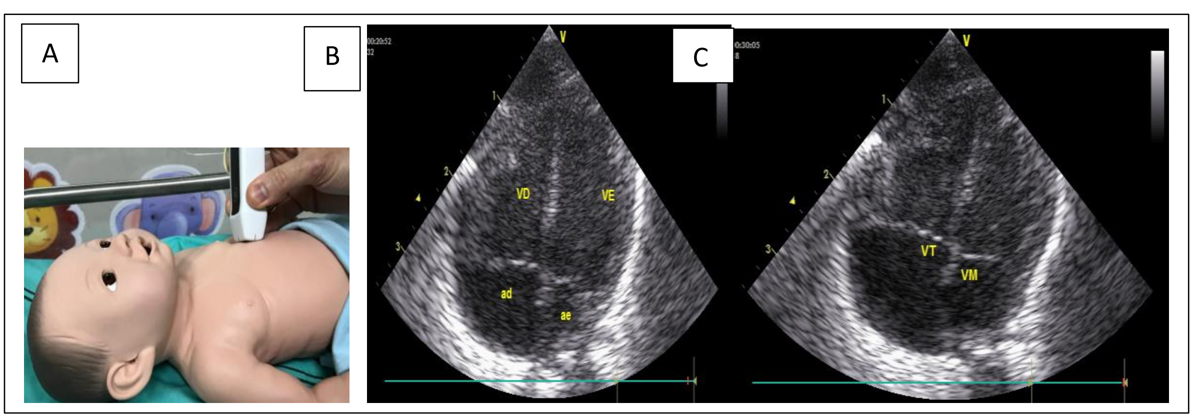

Figure 5: Apical window. A) Transducer positioning, marker facing left. B) Image of the four chambers. C) Observe that the tricuspid valve is a little more inside the right ventricle when compared to the mitral valve and the left ventricle. ad: right atrium, ae: left atrium, RV; right ventricle, LV: left ventricle, MV: mitral valve, VT: tricuspid valve.

In the parasternal long axis window, use color flow to search for ventricular shunt and analyze transmitral flow. This view is ideal for the examination of the interventricular septum for defects, and the origins of the great arteries. Ventricular septal defects may also be visualized here using color Doppler. The aortic valve can be evaluated. In case of valve stenosis, it can be visualized thickened and not "disappear" in systole. Slight inclination of the transducer allows visualization of the tricuspid valve and also the right ventricular outflow tract.[10-12]

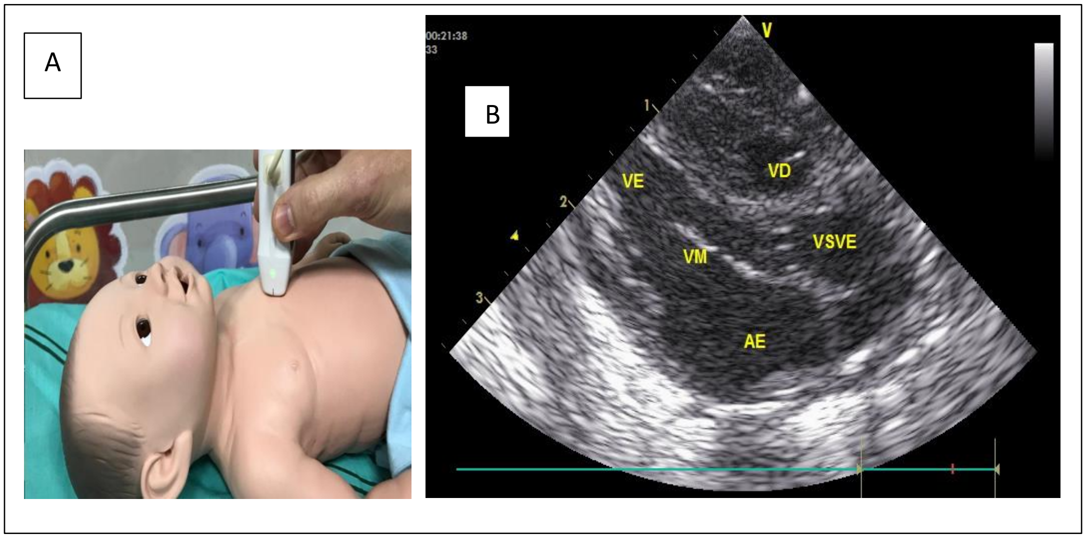

Figure 6: Long axis parasternal window. A) Positioning of the transducer with the marker directed towards the right shoulder. B) Image of the heart chambers. LA: left atrium, RV: right ventricle, LV: left ventricle, MV: mitral valve, LVOT: left ventricular outflow tract.

When moving to the short axis parasternal window, be aware of the existence of interventricular septal deviation (it may be an indirect sign of pulmonary hypertension) and when tilting the transducer, analyze the right ventricular outflow tract. The pulmonary valve can be clearly visualized in this window. Color flow mapping can demonstrate the mosaic pattern common to right ventricular outflow tract obstructions due to high-velocity flow. Measure the flow velocity in the outflow tract and if it is high (above 1.8 m/s) consider the existence of an obstruction in this outflow tract (figure 7). Ideally, go up an intercostal space for a better analysis of the ductus arteriosus and, if present, measure the diameter, flow velocity and direction of flow (figure 8).[9-14]

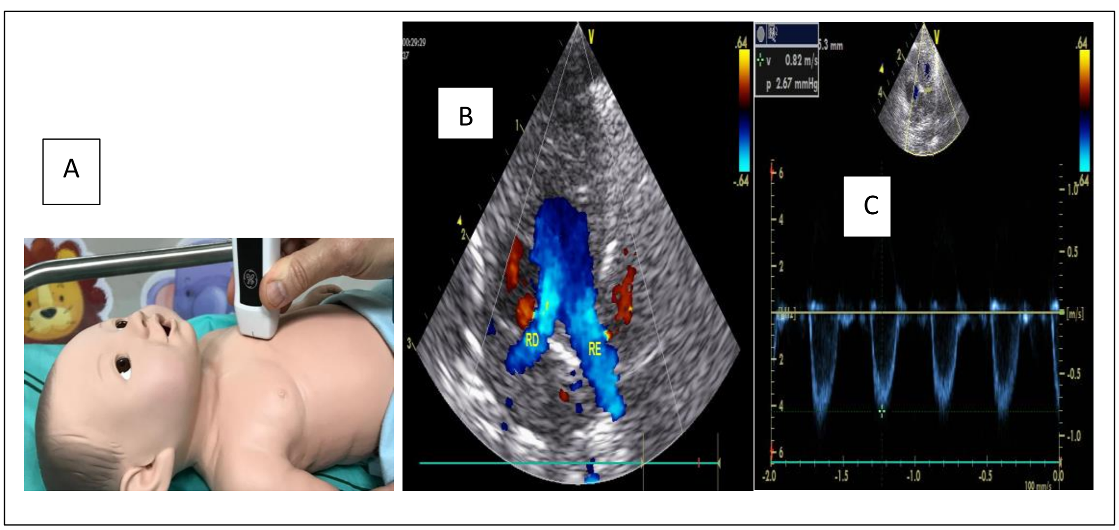

Figure 7: Short axis parasternal window. A) Positioning of the transducer with the marker directed to the left and placing the transducer anteriorly. B) Image of the trunk and branches of the pulmonary artery. C) Analysis of pulmonary artery flow velocity.

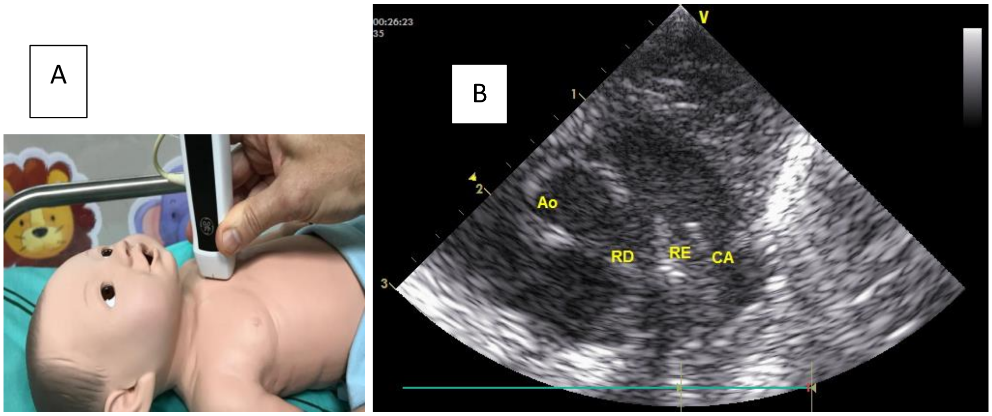

Figure 8: Ductus window, with an intercostal space positioned above the parasternal short axis window. Ao: aorta, RD: right branch of pulmonary artery, RE: left branch of pulmonary artery, CA: ductus arteriosus.

Although the parasternal and suprasternal windows are not always performed in functional echocardiography, it is important to examine them if comprehensive echocardiography has not yet been performed. This parti of the exam allow you to visualize the aortic arch and measure the flow velocity in the descending aorta (figure 9). Its visualization at low speeds (less than 2.4 m/sec) suggests normality. However, be very careful at this point. It is difficult to visualize aortic narrowing, and the velocity is not always high even when there is coarctation of the aorta. If there is a clinical possibility of obstruction of the left ventricular outflow tract, coarctation of the aorta and interruption of the aortic arch cannot be ruled out and specialist examination and/or angio-tomography must be performed. This is also a window to assess whether the pulmonary veins are normally connected to the left atrium.[9-11] Another imaging that is difficult for nonspecialists to perform.

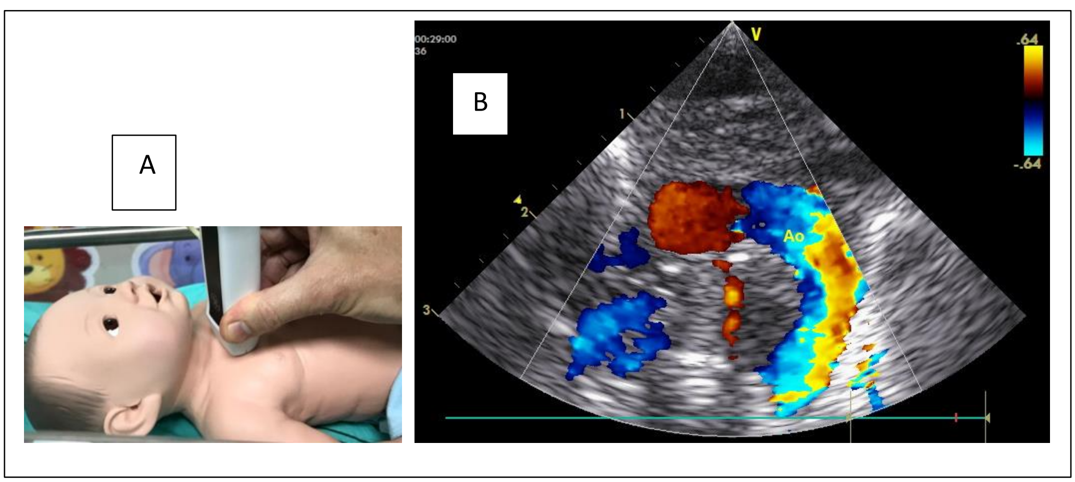

Figure 8: High parasternal window. A) Positioning of the transducer. B) Descending aorta.

In developing countries, the transfer of newborns with suspected heart disease to specialised centres may be difficult and time-consuming. Specialized centers request comprehensive echocardiography confirming heart disease to accept admission. But not all neonatal units have pediatric cardiologists or adult echocardiographers trained to diagnose congenital cardiac disease. In a recent study, we compared the performance of neonatologists performing functional echocardiography and specialists performing comprehensive echocardiography, with good accuracy.[12] The most difficult heart diseases to diagnose are coarctation of the aorta and anomalous pulmonary venous drainage. Thus, imaging non specialists must be aware of clinical data and not rule out the hypothesis of a complex malformation.

European and North American societies draw attention to the difficulty of diagnosing coarctation of the aorta and anomalous pulmonary venous drainage in the neonatal period and have stated that comprehensive echocardiography should initially be performed in case of possibility of congenital heart disease, suggesting that a functional echocardiography be performed after ruling out structural disease.[15-16] They also remember the availability of telemedicine in those necessary cases. Corredera describes in his work that professionals with advanced training in functional echocardiography are unlikely to suspect congenital heart disease if it is present, reaffirming the need for these professionals to recognize their limitations.[2] It is worth emphasizing that point-of-care ultrasonography corresponds to the complement of clinical examination.

Guidelines for the use of point-of-care ultrasound (POCUS) in neonatal and pediatric intensive care endorsed by the European Society of Pediatric and Neonatal Intensive Care have been published to guide clinicians in the use of POCUS for specific indications, although the line between cardiac POCUS and advanced hemodynamic assessment remains somewhat fluid. There is an urgent need to develop cardiac POCUS curriculum and certification to support the widespread and safe use in neonates.[17-18]

The principle of POCUS is the use of ultrasound as a quick and limited bedside tool for diagnostic and procedural applications. In general, training focuses on providing basic ultrasound skills to a wide range of professionals.[6,19] Ideally, neonatologists who perform functional echocardiography should have formal, ongoing training in the search for structural defects. This training should involve theoretical studies and continuous practice, ideally accompanying exams performed by the child cardiologist and participating in the discussion of cases. It is important to highlight the possibility of using telemedicine by sending footage for discussion with a specialist.

4) Conclusion:

The reported cases suggest that non-imaging specialists with adequate training and systematically performing echocardiography can recognize structural defects and diagnose some congenital heart diseases. This professional must be aware of its limitations, as well as perform adequate semiology and not rule out a structural defect based on cardiac ultrasound alone. You can, however, use video of your exam for discussion with a specialist. The diagnosis of congenital heart diseases is not simple and requires a specialist to do it. However, identification of abnormal patterns is useful for managing critically ill patients.

5) Conflict of interest:

The authors have no conflicts of interest to declare.

6) Acknowledgments

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7) Author contributions:

All authors were major contributors on writing the manuscript, conceptualization of work, analyzed and interpreted the patient data, helped in data interpretation and reference checking, conceptualization of work. All authors read and approved the final manuscript.

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