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Review article

Methods for Diagnosing Congenital Anomalies of the Aortic Arch in Children. Advantages and Disadvantages

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Abstract

Congenital heart defects (CHD) account for a third of all congenital malformations and occur in 0.7-1.7% of newborns. Congenital anomalies of the aortic arch (CAAA) are structural abnormalities that affect the development of the aorta and its branches. Congenital anomalies of the aortic arch is very important to diagnose in the neonatal period, as early diagnosis and proper treatment are key factors in improving the long-term prognosis.

The purpose of this review was to study methods for detecting aortic arch abnormalities in infants in order to increase the sensitivity of screening and diagnostic accuracy.

Literature was searched in electronic databases PubMed, MEDLINE, Web of Science, Google Scholar and an electronic library using keywords. The analysis included 20 sources that met the inclusion criteria.

CTA exhibits higher sensitivity in detecting extracardiac structural malformations compared to transthoracic echocardiography, and MRI outpaces transthoracic echocardiography in determining the number of shunts, accurately measures myocardial function, and provides isotropic three-dimensional datasets for complex cardiac abnormalities.

Key words: congenital heart disease, echocardiography, computed tomography, magnetic resonance imaging.

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Introduction

Congenital anomalies of the aortic arch (CAAA) are structural abnormalities that impact the development of the aorta and its branches. There are several types of aortic arch anomalies, including aortic coarctation, interrupted aortic arch, double aortic arch, right aortic arch, and vascular ring. Early diagnosis of aortic arch anomalies is crucial as timely identification helps assess the severity of the defect and select the most appropriate treatment, significantly influencing long-term prognosis [1,2]. Various imaging methods, such as echocardiography (ECHO), magnetic resonance imaging (MRI), and computed tomography (CT), are used for diagnosing this heart defect. These methods accurately determine the location and severity of the anomaly, facilitating the selection of the most effective treatment approach.

The purpose of this review was to explore methods for detecting aortic arch anomalies in children with the goal of enhancing screening sensitivity and diagnostic accuracy.

Search strategy

Literature search was conducted in electronic databases including PubMed, MEDLINE, Web of Science, Google Scholar, and an electronic library using relevant keywords. Seventeen sources meeting the inclusion criteria were analyzed.

Visualization of congenital anomalies of the aortic arch in children

Echocardiography is one of the most readily available and widely utilized primary imaging methods in cardiology, including the diagnosis of aortic arch anomalies in children. One of its main advantages is its high specificity and sensitivity. This method provides high-quality images of the heart and vessels, enabling specialists to accurately identify the location and nature of the anomaly. A review article in "Cardiovascular Ultrasonography" examined the diagnosis of aortic arch anomalies in children using ECHO and noted that ultrasound is the most accessible method for diagnosing aortic arch anomalies, feasible during both pregnancy and after childbirth. The study utilized a series of echocardiographic projections to assess the aortic arch, including three-vessel and tracheal view, short-axis view of the aortic arch, long-axis view of the left aortic arch, and continuous long-axis scanning of the aortic arch. Sensitivity and diagnostic agreement rates of these different images for aortic arch anomalies were analyzed. Results revealed various types of anomalies, such as right aortic arch, double aortic arch, aortic coarctation, and interrupted aortic arch. In most cases, the diagnosis was accurate, with a few misdiagnoses between aortic coarctation and interrupted aortic arch.

The combination of these four projections demonstrated a significant improvement in screening sensitivity and diagnostic agreement. Echocardiography exhibits 100% sensitivity in detecting essential elements of aortic arch anomalies, making it an excellent tool for primary visualization and diagnosis of interrupted aortic arch. However, ultrasound may have limitations in some cases due to restricted visualization, requiring an experienced physician for proper interpretation. Since it is not possible to visualize all anatomical details, surgical intervention should rely on data from computed tomographic angiography or cardiac magnetic resonance imaging, which precisely visualize both abnormal vessels and compressed structures.

The primary advantage of echocardiography is its avoidance of ionizing radiation or intravenous contrast agent use, making it a safe and less invasive method for patients. This is particularly important in the diagnosis of interrupted aortic arch in children, where the risk of adverse effects from alternative imaging methods may be higher. Furthermore, echocardiography can be performed both in utero and after birth, enabling quick and accurate diagnosis of aortic arch anomalies in children. In a review published in 2019, which assessed the accuracy of ECHO in diagnosing interrupted aortic arch in neonates, researchers analyzed data from 115 studies. As a result of the meta-analysis, it was found that ECHO has high diagnostic accuracy in diagnosing aortic arch anomalies in neonates, with a sensitivity of 100% (95% confidence interval 96.5% - 99.1%) and specificity of 99.3% (95% confidence interval 98.5% - 99.7%) [2].

Interruption of aortic arch (IAA) is a congenital heart anomaly that can be challenging to diagnose accurately. Recent developments in echocardiography, particularly three-dimensional echocardiography (3D-ECHO), have shown promise in enhancing the precision and sensitivity of IAA diagnosis. This article explores the advantages of 3D-ECHO compared to traditional two-dimensional echocardiography (2D-ECHO) in the context of IAA diagnosis.

Enhanced Cardiac Visualization: 3D-ECHO enables the creation of more precise three-dimensional images of cardiac structures, including the aorta, pulmonary arteries, and left and right ventricles. This capability allows for a more accurate assessment of heart size, shape, and the localization of defects and anomalies.

Increased Sensitivity: Due to its improved visualization capabilities, 3D-ECHO can detect subtle cardiac structural anomalies that might be missed with conventional 2D-ECHO.

Blood Flow Quantification: 3D-ECHO can be used to quantify blood flow within the heart and blood vessels, particularly valuable for evaluating heart failure and other cardiovascular conditions.

Improved AAI Diagnosis: 3D-ECHO is particularly advantageous in diagnosing IAA because it provides a more precise determination of the defect's location and shape, which can be challenging with 2D-ECHO.

Enhanced Surgical Planning: 3D-ECHO can assist surgeons in planning complex cardiac procedures by creating three-dimensional heart models.

Another study, published in "Cardiology in the Young," investigated the effectiveness of echocardiography in diagnosing IAA. The study included 53 children who underwent echocardiographic examinations. The results demonstrated that echocardiography outperformed chest X-rays in detecting IAA in children. Consequently, the study revealed that transthoracic ECHO has a sensitivity of 100% and a specificity of 98.2% in diagnosing IAA. Despite its advantages, echocardiography has limitations. Some of the drawbacks of ECHO in diagnosing IAA include limited visualization capabilities.

ECHO may have constraints in visualizing specific anatomical structures, especially in cases of complex anomalies or difficulties in obtaining optimal viewing angles. The accuracy and reliability of ECHO results may depend on the experience and qualifications of the performing and interpreting physician. Incorrect scanning techniques or interpretation errors can lead to inaccurate results. Additionally, ECHO may provide incomplete information in some cases, necessitating additional diagnostic methods such as computed tomography (CT) or magnetic resonance imaging (MRI) to achieve a comprehensive diagnosis of IAA.

Computed Tomography (CT) is a powerful tool for diagnosing interruption of aortic arch (IAA) in children. It provides detailed imaging of the chest and aortic anatomy, enabling precise identification of the presence and characteristics of IAA. CT offers several advantages in diagnosing AAI in children: firstly, Solution Imaging: CT provides high-resolution images of chest anatomy and the aorta, allowing for the accurate detection and characterization of AAI. It can identify even small and complex aortic arch anomalies. Secondly, Rapid Execution and Real-Time Scanning: CT allows for swift examination and real-time scanning. This capability is particularly valuable for young children with limited cooperation. The fast scanning process also enables real-time imaging of the heart and aorta, aiding in visualizing anomalies in motion. Thirdly, Assessment of Concomitant Anomalies: CT enables the detection of concomitant anomalies in chest and heart structures that may accompany IAA. This is important to ensure a complete assessment of the patient's condition and planning for surgery, if necessary. The latter is the possibility of 3D visualization; CT provides the ability to create three-dimensional images of the anatomy of the heart and aorta. This allows doctors to study the structures in more detail and plan surgical interventions with high accuracy. Studies conducted over the past 10 years confirm the effectiveness of CT in diagnosing aortic arch break in children. For example, an article published in the journal Monadic Archives Chest Diseases in 2019 presents a retrospective study based on the results of CT angiography in comparison with the results of transthoracic echocardiography of the chest in patients with anomalies of the aortic arch. The study included 203 patients with congenital anomalies, including 107 men and 96 women. The most common aortic arch abnormality was coarctation (19.7%), followed by a right-sided arch with mirror branching (19.2%). Moreover, the most common cardiac abnormalities associated with anomalies of the aortic arch were VSD, LA, and PDA [5].

The sensitivity and specificity of transthoracic echocardiography in the diagnosis of aortic arch abnormalities was 59% and 100% compared to CT angiography. An article published in the journal "Front Pediatric" titled "Accuracy and image quality of wide-detector revolution CT angiography combined with prospective ECG-triggered CT angiography in the diagnosis of congenital aortic arch anomalies in Chinese children" in 2022 explores the effectiveness of using advanced imaging techniques in the diagnosis of complex congenital heart anomalies. Between January 2020 and July 2022, the study examined data from 57 Chinese pediatric patients with confirmed congenital anomalies of the aortic arch who underwent both CT angiography (CTA) using the Revolution CT system and transthoracic echocardiography (TTE) prior to surgery. The study demonstrates the diagnostic accuracy of the combined Revolution wide-detector CT angiography and the prospective ECG trigger approach, demonstrating high sensitivity, specificity, accuracy, positive predictive value, and negative predictive value for the detection of congenital anomalies of the aortic arch. For extracardiac structural malformations, the sensitivity of CTA was 100% and the sensitivity of TTE was 78.6% ($p<0.001$). For intracardiac structural malformations, the sensitivity of CTA was 84.5%, whereas TTE was 92.5% ($p<0.001$). It is noteworthy that the CTA method demonstrates a higher sensitivity in the detection of extracardiac structural malformations compared to TTE. In the field of intracardiac structural abnormalities, TTE is ahead with a slightly higher sensitivity. The study highlights the key role of the Revolution CT system, which boasts a wide detector and uses advanced reconstruction algorithms, offering comprehensive visualization of complex cardiovascular structures. The authors acknowledge the limitations of CTA in the accurate detection of certain intracardiac structural abnormalities, but emphasize its potential as an indispensable tool for preoperative diagnosis and surgical planning. This study makes a valuable contribution to the field of pediatric cardiology by shedding light on the diagnostic utility and image quality achieved through the integration of modern CT angiography techniques. However, the study recognizes its single-center nature and limited sample size, offering opportunities for future research and expanding the results of the study [6,7].

A study published in 2014 in The Egyptian Journal of Radiology and Nuclear Medicine, which aimed to assess the reliability of 64-slice multidetector computed tomography (MDCT) angiography in the preoperative assessment of thoracic aortic coarctation in children, can be considered in favor of the advantages of CT in the diagnosis of aortic arch abnormalities. A total of 24 patients with suspected coarctation of the aorta underwent both Doppler echocardiography and MSCT angiography. The results of MSCT were compared with the results of echocardiography and surgery. The study showed that MSCT angiography has an overall sensitivity of 100% in the diagnosis of extracardiac aortic abnormalities, which is higher than the sensitivity of Doppler echocardiography (92%). However, for the assessment of heart defects, MSCT angiography had an overall sensitivity of 85%, which was lower than the sensitivity of Doppler echocardiography (100%). The study concluded that MSCT angiography with multiplanar and three-dimensional techniques can be considered the method of choice for preoperative assessment of thoracic aortic coarctation in children. One limitation of the study was the small number of patients and the lack of comparison with conventional angiography and magnetic resonance angiography. The authors concluded that MSCT angiography, especially with multiplanar and three-dimensional techniques, is a valuable tool for accurately assessing thoracic aortic coarctation in children, providing important information for preoperative planning and decision-making [8].

Another study published in the Journal of Thoracic and Cardiovascular Surgery in 2016 compared the effectiveness of CT and MRI in diagnosing aortic arch break in newborns. The results showed that CT provides a clearer image of the anatomy and allows for more accurate data on the size of the break and its relationship to the surrounding structures.

The main disadvantage of CT, especially in the pediatric population, was the cumulative radiation dose from repeated examinations in the interventional period after surgery or endovascular treatment. However, with the advent of new modern scanners, it is possible to achieve a dose reduction of up to 90% in routine studies without losing diagnostic accuracy [20]. For example, the use of low-dose radiation techniques, such as adaptive dose reduction protocols, can be applied to reduce the risks associated with radiation [6]. In recent years, efforts have been made to develop scanning protocols that allow for a reduction in the radiation dose of CT scans and maintain high diagnostic accuracy, and this has necessitated the introduction of an imaging technique such as magnetic resonance imaging (MRI).

The benefits of **magnetic resonance imaging** in diagnosing aortic arch interruption in children include: No use of radiation: MRI does not use X-rays, making it safe for use in children. Instead, MRI uses magnetic fields and radio waves to create detailed images of the anatomy of the chest and aorta. High resolution: MRI provides high image resolution, which allows you to visualize in detail abnormalities in the structure of the aortic arch. This allows you to accurately determine the location and characteristics of the break. Blood flow assessment: MRI can be used to assess blood flow in the aorta and vessels. This allows you to determine the presence of stenosis or regurgitation, as well as assess the overall function of the heart and aorta. 3D imaging capability: MRI allows you to create three-dimensional images of the anatomy of the heart and aorta. This allows for a more detailed study of abnormalities and helps in planning surgery, if necessary. However, it is worth noting that MRI may be limited in availability and require a longer scan time. Some children, especially infants and children with limited cooperation may find it difficult to remain still during an MRI session. An article published in the journal Cardiology in the Young discusses a rare and complex case of congenital heart disease with an aortopulmonary window in combination with an interrupted aortic arch observed in a premature baby. This particular case was distinguished by the successful use of a high-field open MRI system with a field of 1.0 Tesla, which provided excellent image resolution and allowed the safe examination of an intubated newborn. The successful use of cardiac MRI in the case of an extremely premature baby weighing only 1.7 kg suggests that this imaging technique can be a feasible and effective universal study for the evaluation of complex congenital heart defects even in very young newborns [9].

The strengths of MRI include the identification of high-resolution extra cardiac anatomical structures, such as large arteries, systemic and pulmonary veins, as well as the assessment of blood flow in vessels and valves. This method also determines the number of shunts, accurately measures myocardial function, and provides isotropic three-dimensional datasets for complex cardiac abnormalities. It is important to note that MRI achieves this without ionizing radiation, making it a valuable tool, especially for patients who do not have sufficient clinical or echocardiographic data. Also, when diagnosing an anomaly of the aortic arch, MRI is preferable due to multilane imaging, that is, it allows you to obtain images in various planes, including transverse, longitudinal and coronal sections. This provides a complete assessment of the aortic arch break anomaly and helps in determining its exact location and shape. It also has a high tissue contrast, since MRI has a high contrast between different tissues, which allows for better visualization of abnormalities in the structure of the aortic arch and surrounding tissues. This is especially important when detecting concomitant anomalies. It should also be emphasized that it is excellent for visualizing extra cardiac structures, including the main arteries and veins, with high spatial resolution. MRI can also assess vascular and volvuli blood flows, quantify shunts, and accurately measure myocardial function, regardless of ventricular morphology [10].

An article published in the American Journal of Roentgen ology in 2015 summarized the role of CT and MRI in the diagnosis of aortic arch abnormalities. MSCT has the highest resolution among non-invasive imaging techniques used to assess the cardiovascular system and aorta. With isotropic reformatted images created in several different planes, MSCT can provide excellent anatomical detail of the aorta and coarctation segment, as well as other related aortic and cardiac abnormalities. With ECG synchronization, it is possible to evaluate the heart in great detail using the same contrast bolus to identify additional potential birth defects. MR angiography (MRA) has become widespread over the past two decades for non-invasive assessment of the heart and blood vessels. ECG-synced T1-weighted images with double inversion of black blood are particularly useful for the anatomical details of the coarctation segment and other adjacent anatomical structures [13].

MRA is a reliable method for evaluating the aorta that allows you to obtain excellent anatomical details. MRA images can be obtained with or without intravenous gadolinium-based contrast agents. Time-of-flight MRA can be performed without intravenous contrast in patients with chronic kidney disease. Contrast-enhanced MRA is the preferred method for anatomical evaluation in patients without known kidney problems. In addition to anatomical imaging, functional information can also be obtained using phase-contrast imaging. Phase contrast sequences and established free precession (SSFP) are useful for objective and subjective assessment of the pressure gradient in the stenotic segment, which is an important parameter for planning surgical and endovascular intervention [11]. Thoracic aortic blood flow volumes measured below and above the level of coarctation can also help quantify the extent and severity of collateral blood flow in a non-invasive way. The combination of morphological and functional data collected by MRI has excellent sensitivity (95%) and specificity (82%) for assessing aortic coarctation. Aortic MRA with functional information is reliable and can be performed in 10-20 minutes [12].

Echocardiography provides 100% sensitivity in detecting elements of an aortic arch anomaly and serves as a valuable primary imaging technique. With the help of the development of medical technology, echocardiographic projections are used to assess abnormalities, demonstrating improved screening sensitivity and diagnostic compliance [18]. The possibilities of 3D echocardiography, which provides more accurate visualization of heart structures, increased sensitivity, determination of blood flow volume, and improved diagnosis of aortic arch anomaly, are also being investigated [14]. CTA exhibits higher sensitivity in detecting extracardiac structural malformations compared to TTE, and MRI outpaces TTE in determining the number of shunts, accurately measures myocardial function, and provides isotropic three-dimensional datasets for complex cardiac abnormalities.

CT scans pay special attention to high-resolution imaging, speed of execution, and the ability to assess concomitant abnormalities [15]. MRI is particularly valuable in complex cases and provides detailed anatomical and physiological information. It is considered an increasingly important tool in the diagnosis of congenital heart defects in children and provides isotropic three-dimensional datasets for complex cardiac abnormalities.

The specificity and sensitivity of echocardiography provide a detailed understanding of the anatomy and localization of the aortic arch anomaly, facilitated by various projections such as three-vessel and tracheal, short aortic arch, long axis of the left aortic arch, and continuous projections of aortic arch scanning [16]. Echocardiography provides 100% sensitivity in detecting elements of the aortic arch anomaly and serves as a valuable primary imaging method, especially when the aortic arch is interrupted. While limited images and expert interpretation may be limitations, the non-invasive nature of the method, devoid of radiation or contrast agents, ensures patient safety. The main advantages of CT include its ability to detect even complex abnormalities, fast execution suitable for children who refuse to stay on the CT table for long periods of time, real-time imaging to assess heart and aortic movements, detection of concomitant abnormalities, and 3D imaging for accurate surgical planning [17-20].

Conclusions

Notably, the combination of these methods improves screening sensitivity and diagnostic matches. MRI offers many advantages, including no radiation, high image resolution, the ability to assess blood flow, and three-dimensional imaging capabilities. Research highlights the potential of MRI as a feasible and effective method for assessing complex congenital heart defects even in very young newborns, indicating its growing importance in the medical field. The widespread availability of MRI scanners and the growing medical expertise contribute further to its role in the treatment of such conditions by providing comprehensive anatomical and physiological information.

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Балалардағы аорта доғасының туа біткен ауытқуларын визуализациялау әдістері. Артықшылықтары мен кемшіліктері

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Түйіндеме

Туа біткен жүрек ақаулары (ЖЖА) барлық туа біткен ақаулардың үштен бірін құрайды және жаңа туған нәрестелердің 0,7-1,7% құрайды. Аорта доғасының туа біткен аномалиялары - аортаның және оның тармақтарының дамуына әсер ететін құрылымдық ауытқулар. Аорта доғасының туа біткен аномалиялары неонатальды кезеңде диагностикалау үшін өте маңызды, өйткені ерте диагностика және дұрыс емдеу ұзақ мерзімді болжамды жақсартудың негізгі факторлары болып табылады.

Бұл зерттеудің мақсаты скринингтің сезімталдығын және диагностикалық дәлдікті арттыру үшін нәрестелердегі аорта доғасының ауытқуларын анықтау әдістерін зерттеу болды.

Әдебиеттер PubMed, MEDLINE, Web of Science, Google Scholar электронды дерекқорларында және кілт сөздерді пайдаланып электронды кітапханада іздестірілді. Талдау қосу критерийлеріне сәйкес келетін 20 дереккөзді қамтыды.

Компьютерлік томографиялық ангиография жүректен тыс құрылымдық ақауларды анықтауда трансторакальды эхокардиографиямен салыстырғанда жоғары сезімталдық танытатынын және магниттi-резонансты томография шунттар санын анықтауда трансторакальды эхокардиографиядан асып түсетінін, миокард қызметін дәл өлшейтінін және күрделі жүрек ақаулары үшін изотропты үш өлшемді деректер жиынтығын беретінін анықтадық.

Түйін сөздер: жүректің туа біткен ақауы, эхокардиография, компьютерлік томография, магниттi-резонансты томография.

Методы визуализации врожденных аномалии дуги аорты у детей. Преимущества и недостатки

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Резюме

Врожденные пороки сердца (ВПС) составляют треть всех врожденных пороков развития и встречаются у 0,7-1,7% новорожденных. Врожденные аномалии дуги аорты - это структурные аномалии, влияющие на развитие аорты и ее ветвей. Врожденные аномалии дуги аорты очень важно диагностировать в неонатальном периоде, так как ранняя диагностика и надлежащее лечение являются ключевыми факторами улучшения долгосрочного прогноза.

Целью данного обзора было изучение методов выявления аномалии дуги аорты у младенцев с целью повышения чувствительности скрининга и точности диагностики.

Поиск литературы осуществлялся в электронных базах данных PubMed, MEDLINE, Web of Science, Google Scholar и электронной библиотеке с использованием ключевых слов. В анализ были включены 20 источников, которые соответствовали критериям включения.

Компьютерная томографическая ангиография демонстрирует более высокую чувствительность при выявлении экстракардиальных структурных мал формаций по сравнению с трансторакальной эхокардиографией, а магнитно-резонансной томография опережает трансторакальную эхокардиографию в определений количество шунтов, точно измеряет функцию миокарда и предоставляет изотропные трехмерные наборы данных для сложных сердечных аномалий.

Ключевые слова: врожденный порок сердца, эхокардиография, компьютерная томография, магнитно-резонансная томография.