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Beyond stethoscopes: Advancing Diagnostic Precision in Pediatric Cardiology A narrative review

Más allá de los estetoscopios: avances en la precisión diagnóstica en cardiología pediátrica Una revisión narrativa

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ABSTRACT

Auscultation, a diagnostic method with a rich history dating back to the time of Hippocrates, has long been a fundamental approach to identifying pediatric heart diseases. Despite its historical significance, the challenge of distinguishing innocent murmurs from those indicating structural heart defects in neonates persists, with conditions such as aortic stenosis, pulmonary stenosis, and atrial septal defect potentially being misinterpreted as innocent murmurs, highlighting the limitations of traditional methods. Although cardiac auscultation remains a cost-effective pre-screening tool, its effectiveness is further hindered by the subjectivity and expertise required for interpretation. A global comparison of auscultation skills among internal medicine trainees revealed suboptimal performance, emphasizing the need for innovative solutions. The integration of technology and Artificial Intelligence (AI) in pediatric cardiology offers a promising avenue for enhancing diagnostic accuracy and patient care. This paper explores the transformative impact of technology and AI in diagnostic cardiology addressing the limitations of traditional auscultation.

Key Words: artificial intelligence, echocardiography, heart auscultation, heart disease, heart sound analysis.

RESUMEN

La auscultación, un método de diagnóstico con una rica historia que se remonta a la época de Hipócrates, ha sido durante mucho tiempo un enfoque fundamental para identificar enfermedades cardíacas pediátricas. A pesar de su importancia histórica, el desafío de distinguir los soplos inocentes de los que indican defectos cardíacos estructurales en los neonatos persiste, y afecciones como la estenosis aórtica, la estenosis pulmonar

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y el defecto del tabique auricular pueden malinterpretarse como soplos inocentes, lo que resalta las limitaciones de los métodos tradicionales. Si bien la auscultación cardíaca sigue siendo una herramienta de preselección rentable, su eficacia se ve obstaculizada aún más por la subjetividad y la experiencia requeridas para la interpretación. Una comparación global de las habilidades de auscultación entre los residentes de medicina interna reveló un desempeño subóptimo, lo que enfatiza la necesidad de soluciones innovadoras. La integración de la tecnología y la inteligencia artificial (IA) en cardiología pediátrica ofrece una vía prometedora para mejorar la precisión del diagnóstico y la atención al paciente. Este artículo explora el impacto transformador de la tecnología y la IA en la cardiología diagnóstica abordando las limitaciones de la auscultación tradicional.

Palabras Claves: inteligencia artificial, ecocardiografía, auscultación cardíaca, enfermedades cardíacas, análisis de los sonidos cardíacos.

Introduction

Auscultation, a long-standing and essential method for detecting heart diseases, traces its origins back to the ancient Greek physician, Hippocrates. In the 19th century, physicians employed direct ear-to-chest contact for screening. However, in 1816, René Laennec revolutionized this practice with the invention of a 31 cm-long cylindrical wooden stethoscope¹. Further modifications, such as the incorporation of rubber tubing in 1829, culminated in the contemporary stethoscope design established in 1926.

In more than 50% of children and adolescents, auscultation reveals innocent heart murmurs, with the highest frequency observed between the ages of 3-6 years and 8-12 years². A murmur is an abnormal sound arising from turbulent blood flow, which occurs due to increased flow or structural heart defects that disrupt the normal harmonious flow of blood. This phenomenon is usually absent in normal vascular conditions, where blood flow is smooth and silent. These are further classified into innocent and pathological murmurs. Innocent murmurs are asymptomatic and occur in anatomically and physiologically normal hearts. The diverse nature of murmurs, in terms of timing, duration, intensity, pitch, and shape, is linked to various structural heart diseases, enabling their identification and understanding^{3,4,5}.

Organic murmurs, associated with conditions such as aortic stenosis, pulmonary stenosis, and atrial septal defect (ASD), can be mistaken for innocent murmurs. Distinguishing between these types demands a high level of expertise in cardiac auscultation, as other non-cardiac conditions, such as supraclavicular or carotid murmurs, may mimic the murmur of aortic stenosis, coarctation, carotid stenosis, or subclavian artery stenosis.

Cardiac murmurs offer invaluable insights into cardiovascular pathologies. While cardiac auscultation is a widely accessible method, its effectiveness relies on expert interpretation, posing challenges in resource-constrained settings. Limited access to clinical experts and infrastructure hinders widespread screening and management of cardiac diseases in such environments.

Is there a need for improved auscultation in pediatric population?

Congenital heart anomalies (CHA) pose a significant challenge in pediatric healthcare, occurring at an incidence rate of 0.8 to 1% per 1000 live births^{6,7}. Although traditional screening methods, such as second-trimester ultrasonography and postnatal clinical examination, are used, their detection rates for CHA are limited, resulting in undiagnosed cases, particularly with the trend of early hospital discharge. It is estimated that over half of children with CHA remain undiagnosed despite routine clinical examinations^{8,9,10}. A significant proportion of neonates with audible murmurs in the neonatal period have structural heart defects, making it difficult to distinguish innocent from organic heart murmurs based on auscultative characteristics¹¹. This distinction is essential for healthcare professionals involved in heart disease screening.

In a study involving over 900 children with innocent-sounding murmurs at a pediatric cardiology clinic, abnormal findings in medical history, physical examination, or diagnostic tests showed 67% sensitivity but only 38% specificity for detecting structural heart lesions in infants under six weeks. Sensitivity increased to 100% in infants older than six weeks, but specificity decreased to 28%. While this information is helpful in ruling out structural causes of an innocent murmur in older infants and children, it lacks utility in younger infants¹². A Norwegian study revealed that only 10% of children referred to a cardiac center for investigating a cardiac murmur were diagnosed with a congenital cardiac lesion, with 71% of referrals made by general practitioners, and only 17% providing a diagnosis¹³. The diagnosis of an innocent heart murmur in children and adolescents is based on specific criteria, including the absence of abnormal physical examination findings (except the murmur), a negative review of systems, a history without risk-increasing features for structural heart disease, and distinctive auscultatory characteristics^{14,15}. These criteria do not apply to newborns or infants under one year of age, as a higher prevalence of asymptomatic structural heart disease exists within this population¹⁶. It is recommended that, in situations where the classification of an innocent murmur is unclear, a referral for echocardiography or to a pediatric cardiologist be considered.

Pediatric cardiologists exhibit a higher level of accuracy in identifying structural heart defects in infants and children presenting with heart murmurs; their sensitivity in detecting pathological heart murmurs in newborns ranges from 80.5% to 94.9%, with specificity varying between 25% and 92%¹⁷. The accuracy of a pediatric cardiologist in identifying pathological murmurs depends on several factors, including diagnostic confidence. In newborns presenting with a heart murmur, it may be deemed unneces-

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sary to perform an echocardiography if a pediatric cardiologist can confidently diagnose an innocent murmur¹⁸. Nonetheless, considering the relatively high prevalence of structural heart disease in asymptomatic newborns with murmurs, further diagnostic measures should be considered.

This underscores the urgent need to enhance medical practitioners' ability to identify murmurs indicative of structural heart disease. Having received FDA approval for aiding physicians in detecting abnormal heart murmurs, Computer-aided auscultation (CAA) emerges as a potential solution to this dilemma.

Is digital technology required in auscultation devices?

Cardiac auscultation with stethoscopes remains a widely used and cost-effective method for cardiac pre-screening. However, its diagnostic sensitivity and accuracy are limited due to the expertise and experience required for accurate interpretation, and the data acquired through auscultation is subjective and lacks a permanent, objective record. This information is challenging to replicate among different examiners, leading to substantial disagreement among medical professionals^{19,20,21}. Furthermore, inadequate training of healthcare professionals in cardiac auscultation has been raised as a concern over the past decade. Despite the potential cost-effectiveness of cardiac auscultation, both students and clinicians often demonstrate incompetence in performing it effectively²². A research study was conducted with 314 senior internal medicine and family practice residents from diverse training programs across the United States. The findings revealed that only 20% of abnormal heart sounds were accurately identified from auscultatory tapes. Although residents who received formal auscultatory training demonstrated greater confidence, their accuracy did not significantly increase²³. Moreover, a comparison of auscultation skills among internal medicine trainees in the U.S., Canada, and Britain indicated suboptimal performance in all three countries, with slightly better results in Canada²⁴.

Therefore, the development of tools for automated classification of specific murmur types is necessary and clinically significant. While experienced cardiologists can successfully distinguish specific heart sound patterns, young and inexperienced physicians face challenges in making accurate diagnoses through auscultation.

Recent innovation in cardiac diagnostics

Echocardiography

In light of the limitations associated with auscultation, phonocardiography was developed. The visual representation of acoustic data allowed for a more accurate assessment of the timing and acoustic characteristics of heart sounds and murmurs compared to traditional auscultation²⁵. Echocardiography has experienced significant advancements since its inception over 30 years ago, when Keidel pioneered the use of ultrasound to explore the heart²⁶. The introduction of two-dimensional echocardiography significantly revolutionized the field, enabling real-time and cross-sectional imaging. The transition from M-mode to two-dimensional imaging, coupled with the integration of Doppler and color flow, has established echocardiography as an indispensable diagnostic tool in cardiology²⁷. The progression towards three-dimensional (3D) imaging represents a notable development, providing real-time volumetric imaging and enhancing accuracy in evaluating cardiac chamber volumes. This technology offers comprehensive views of cardiac structures, facilitating surgical interventions and postoperative assessments. The ongoing refinement of 3D imaging ensures its integration into routine clinical practice²⁸.

While echocardiography offers numerous benefits, primary care physicians should also be cognizant of its limitations. Approximately 5% to 10% of studies may be insufficient for interpretation due to patient demographics and the echocardiography lab's experience. Additionally, operator dependence is a concern, affecting data acquisition and interpretation. Reproducibility can be challenging, and there are no standard criteria for age-related valve changes. The qualitative and subjective nature of color flow imaging in valvular regurgitation grading adds to these concerns, although quantitative techniques are emerging. It is also worth noting that diagnostic errors are common in pediatric echocardiography conducted in community-based adult labs²⁹.

Acoustic cardiography

The acoustic cardiography technology, which is a more recent advancement, enables the simultaneous acquisition of both electrocardiogram (ECG) and cardiac acoustical data. This user-friendly and cost-effective approach allows for a detailed assessment of the left ventricular function during systole and diastole. The present system offers a computerized analysis and graphical portrayal of its findings. Its medical applications extend to the evaluation of illnesses such as heart failure, ischemia, cardiac arrhythmias, and the optimization of cardiovascular medication and equipment therapies³⁰.

The application of acoustic cardiography, which utilizes digital data for automated interpretation, eliminates the necessity for specialized expertise in heart sound or ECG data analysis. By employing standardized sensor locations, uniform filtering, and processing, this approach surpasses the labor-intensive nature of traditional phonocardiography. The Audicor® technology allows for continuous recording of concurrent sound and ECG data, facilitating cardiovascular monitoring and optimizing cardiac synchronization therapy devices. Studies have demonstrated its efficacy in assessing systolic and diastolic function in heart failure patients, as well as in monitoring patients undergoing cardiotoxic chemotherapy 31. Additionally, its early detection capability for acutely decompensated heart failure in emergency units has been showcased by Collins et al^{32,33}. The cost-effectiveness and user-friendly nature of acoustic cardiography make it a practical tool for mass screening, particularly in regions with prevalent diseases such as rheumatic heart disease³⁴. Overall, acoustic cardiography emerges as a reliable and cost-effective alternative to echocardiographic methods, demonstrating comparable efficacy to invasive cardiac catheterization and noninvasive echocardiography. By seamlessly integrating with routine ECG testing, it swiftly addresses the unmet clinical need for expedited and accurate diagnoses in emergency settings.

Artificial Intelligence and Machine Learning

Artificial intelligence has become increasingly prevalent in the field of computer-aided diagnosis in recent years. Artificial Intelligence (AI) manifests as a type of intelligence displayed by devices, emulating human cognitive functions such as learning and problem-solving. Machine Learning (ML), a subset of AI, involves creating and training mathematical models using extensive datasets. The healthcare sector has increasingly integrated ML, utilizing algorithmic advancements and the abundance of "big data" to enhance diagnostics, improve test reliability, reduce errors related to cognitive bias, engage patients, and streamline administration³⁵. In cardiovascular medicine, ML applications have expanded to include conditions like heart failure, cardiomyopathy, hypertension, and coronary artery disease, with recent focus on mitral and tricuspid valve disorders^{36,37,38}.

Particularly noteworthy is the progress made in detecting and classifying heart sounds using artificial neural networks (ANNs) and deep neural networks (DNNs). Jou-Kou Wang et al. proposed a novel algorithm, the temporal attentive pooling-convolutional recurrent neural network (TAP-CRNN) model, for automatically identifying systolic murmurs in patients with ventricular septal defects (VSD)³⁹.

In the field of medical imaging, a significant challenge is the reliance on skilled operators for tasks such as image acquisition, interpretation, and decision-making. Artificial Intelligence (AI) presents a transformative solution, utilizing Machine Learning (ML) to acquire expertise in rule learning and pattern recognition from diverse datasets. These datasets include essential factors like pixel density, brightness, vector movement, and measurements. Segmentation allows for the division of images or volumes into landmarks, facilitating automated measurements of 2D dimensions or Doppler velocities, thereby enhancing reproducibility and efficiency^{40,41}. Furthermore, the implementation of deep learning algorithms reduces the reliance on highly trained individuals, offering automated analysis of chamber volumes and function⁴². Additionally, Al's ability to facilitate remote training enables skill development without the need for in-person contact, which is particularly beneficial in underserved communities⁴³.

Super Stethoscopes

Furthermore, Shimpei et al.⁴⁴ introduced the Super StethoScope, a device designed to capture and record both electrocardiographic and heart sounds, which facilitates the detection of heart rate variability and enhances the signal-to-noise ratio in the audible frequency range, while also capturing heart sounds across both audible and inaudible frequency ranges. This innovative device enabled the visualization of quantitative results, ensuring precise data interpretation during remote auscultations, while mitigating potential disruptions arising from fluctuations in sound quality.

The use of digital stethoscopes has the potential to significantly enhance the detection of murmurs through the conversion of acoustic sounds into electronic signals, which can then be amplified, filtered, and digitalized. This technology, when combined with advanced analysis software, has the potential to transform auscultation into a more objective and quantitative tool for clinical heart evaluation. This innovation has the potential to enhance the assessment of innocent murmurs, mitigate the variability resulting from human acoustic limitations, and improve the teaching of cardiac auscultation⁴⁵. The integration of digital stethoscopes, which offer features such as sound recording, adjustable playback speeds, visual displays, and database creation, presents a unique opportunity to improve the efficiency of auscultation instruction. Additionally, in regions such as sub-Saharan Africa, where there is a shortage of trained specialists in healthcare, the use of technology to support community health workers may be a potential solution to this problem. The implementation of Computer-Aided Auscultation (CAA) in educational programs could represent an important step in addressing these challenges and improving healthcare outcomes⁴⁶.

Conclusion

In summary, the progression of computer and AI-based auscultation in pediatric cardiology offers a promising prospect for enhancing diagnostic accuracy and patient care. To capitalize on this potential, it is essential for healthcare professionals, researchers, and policymakers to actively support and incorporate these technologies into everyday practice. This includes providing ongoing education on AI implementation, nurturing collaboration between clinicians and technology experts, and establishing strict regulatory frameworks. The urgency of transitioning from conventional methods to advanced, technology-driven approaches is crucial in ensuring improved cardiac care for pediatric patients. However, only the future will determine whether the binaural stethoscope will become a relic of the past, like its predecessor, the monaural stethoscope, or will remain a relevant and valuable clinical diagnostic tool.

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