Artificial intelligence: how technology helps in the diagnosis of congenital heart disease

Inteligência artificial: como a tecnologia auxilia no diagnóstico das cardiopatias congênitas

Inteligencia artificial: cómo la tecnología ayuda en el diagnóstico de cardiopatías congénitas

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RESUMO

Objetivo: A cardiopatia congênita (CC) é o defeito congênito mais recorrente, que apresenta uma baixa taxa de detecção precoce. Em tal cenário a IA se mostra promissora, sobretudo no que tange impactos clínico-radiológicos da ecocardiografia fetal. Esta revisão de literatura sistemática e qualitativa foi elaborada mediante a pesquisa bibliográfica nas bases de dados Biblioteca Virtual da Saúde (BVS) e PubMed. Embora a IA auxilie na detecção de CC e contribua com profissionais menos experientes, observa-se limitações e sua implementação requer validação clínica. O impacto na redução de subdiagnósticos e na melhora de desfechos clínicos é limitado, depende do auxílio humano e carece de estudos para comprovar o impacto na ecocardiografia fetal.

Palavras-chave: Cuidado Pré-natal; Defeitos cardíacos congênitos; Inteligência artificial.

ABSTRACT

Objective: Congenital heart disease (CHD) is the most common birth defect, which has a low rate of early detection. In such a scenario, AI shows promise, especially regarding the clinical-radiological impacts of fetal echocardiography. This systematic and qualitative literature review was prepared through bibliographic research in the Virtual Health Library (VHL) and PubMed databases. Although AI helps in the detection of CC and contributes to less experienced professionals, limitations are observed and its implementation requires clinical validation. The impact on reducing underdiagnosis and improving clinical outcomes is limited, depends on human assistance, and lacks studies to demonstrate the impact on fetal echocardiography.

Keywords: Heart Defects; Generative Artificial Intelligence; Prenatal Care.

RESUMEN

Objetivo: La enfermedad cardíaca congénita (EC) es el defecto congénito más común y tiene una baja tasa de detección temprana. En este escenario, la IA muestra resultados prometedores, especialmente en lo que respecta a los impactos clínico-radiológicos de la ecocardiografía fetal. Esta revisión sistemática y cualitativa de la literatura se elaboró a través de la investigación bibliográfica en las bases de datos de la Biblioteca Virtual en Salud (BVS) y PubMed. Si bien la IA ayuda en la detección del CC y aporta a los profesionales menos experimentados, se observan limitaciones y su implementación requiere validación clínica. El impacto en la reducción del subdiagnóstico y la mejora de los resultados clínicos es limitado, depende de la asistencia humana y carece de estudios que demuestren el impacto en la ecocardiografía fetal.

REVISÃO

Descriptores: Cardiopatías Congénitas; Inteligencia Artificial Generativa; Atención Prenatal

Introduction

Congenital heart defects (CHDs) are a set of diseases characterized by the presence of a congenital anomaly, being one of the main pathologies responsible for high infant morbidity and mortality worldwide. Thus, diagnosis during the gestational period allows for assertive neonatal management, as well as the refinement of family guidance.[1]

CHD is a recurrent congenital defect, affecting between 0.6 and 1.2% of live births. Children in fetal age affected may have more than one CHD associated, the most common being ventricular septal defect (VSD), atrioventricular septal defect (AVSD), Tetralogy of Fallot (TOF), and transposition of the great arteries (d-TGA). Such pathologies can manifest through abnormalities in the cardiac structure or function, which are already evident in the fetal period.[2]

Some adversities permeate the prenatal detection rate, such as: the diminished cardiac size and its imprecise anatomy; the generation of fetal movements with the application of the probe; the rapidity of fetal heartbeats; and the unpreparedness of professionals related to fetal echocardiography.[1] It is observed that 9 out of 10 fetal heart diseases do not manifest clinically, which hinders early recognition in prenatal care.[2]

Currently, fetal echocardiography demonstrates a sensitivity of 68.5% for the detection of CHD, and, in the case of low-risk pregnant women, this number represents 36.1%. About 25% of newborns with unidentified CHD are discharged from the hospital without the diagnosis of the disease. AI, through the use of advanced algorithms and investigation and imaging technologies, influences the clinical-radiological scope. Since its use in detection is promising regarding the mitigation of the reduced number of early diagnoses of CHD.[3]

Method

This study consists of a systematic literature review, of a qualitative nature, carried out through a bibliographic survey in free-access databases: Virtual Health Library (VHL) and PubMed.

For the selection of publications, the descriptors: “artificial intelligence,” “congenital heart defects,” and “prenatal,” separated by the Boolean operator AND, all registered in the Health Sciences Descriptors (DeCS), were used. The inclusion criteria were: full-text articles, in Portuguese and English, published in the last 5 years. This resulted in 54 articles in PubMed and 30 in VHL. Publications that did not meet the inclusion criteria were disregarded, and in the end, 9 articles were adopted.

Results and Discussion

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The current model of congenital heart defect screening X approaches based on artificial intelligence

Congenital heart disease (CHD) is one of the causes of neonatal mortality, with the incidence of CHD in live births being 0.8 to 1.1% worldwide.[4] The prenatal diagnosis of fetal CHD is associated with reduced morbidity and mortality after birth.[5] Furthermore, the prenatal detection of CHD favors a promising postnatal prognosis for some forms of congenital heart disease and, in some situations, may allow therapeutic intervention while the fetus is still *in utero*.[6]

From this perspective, CHD contributes to 30% of infant mortality due to congenital malformations.[7] Currently, through the action of experienced sonographers, most of the main CHDs can be diagnosed in the second trimester of pregnancy, and approximately two-thirds of them can be detected before 14 weeks of gestation. However, the diagnosis of CHD remains below the ideal due to anatomical complexity, cardiac function, and the inherent difficulty of ultrasound examination in the fetal heart.[8]

AI, combined with the fetal echocardiogram exam, has been studied to improve the accuracy of fetal congenital heart defect diagnosis.[6] In this context, AI is the discipline that makes machines act like human intelligence, and with its rapid expansion, numerous algorithms are used in the field of medical imaging.[9] In this context, AI is the discipline that makes machines act like human intelligence, and with its rapid expansion, numerous algorithms are used in the field of medical imaging.[10]

Fetal echocardiography, an exam that evaluates the fetal heart, is performed between the 18th and 24th weeks of pregnancy. However, complex anatomical structures, rapid fetal heartbeats, body movements, and the thickness of the maternal abdominal wall require advanced experience from sonographers to obtain stable images and analyses. With the aid of AI, these professionals can be guided to make diagnoses, identify important structures, locate lesions, and classify diseases with more assertiveness.[9]

In this context, an American study involving AI measurements in screening images created results 20 to 40% better than current detection rates, comparable to the predictions made by a pre-selected group of specialists. These comparable results can be attributed to the precise measurements performed by AI and the extensive data set analyzed, in addition to the effective feature selection process performed by the study.[7]

Still, from this perspective, another study carried out in the United Kingdom found that AI proved promising in the automatic classification of images suggestive of congenital heart disease on ultrasound. The study in question dealt with the prenatal diagnosis of hypoplastic left heart syndrome (a type of CHD) by ultrasound using artificial intelligence and found that the AI model used was highly sensitive and would detect 10 additional cases of hypoplastic left heart syndrome per year. In contrast, the specificity of the model using artificial intelligence was inferior compared to the current screening system, which means that the traditional model generates fewer false positives than the one that uses AI assistance.[6]

Challenges of ultrasound diagnosis and the potential of artificial intelligence in improving prenatal detection

The fetal heart is, possibly, the most difficult organ to evaluate, so routine anomaly scanning becomes complex. Thus, the small fetal heart, maternal weight, amniotic fluid volume, and even fetal position impose greater demands on sonographers and equipment to obtain the best performance. The accurate prenatal diagnosis of CHD allows physicians to provide appropriate consultation and treatment, which improves the short- and long-term prognosis of fetuses.[9]

4D ultrasound enhanced by AI provides better fetal images, allowing for the detection of anomalies earlier when compared to traditional 2D images. Deep learning algorithms identify patterns in large datasets and increase diagnostic accuracy. Thus, the integration of AI increases the capacity of 4D ultrasound to identify fetal anomalies, placental dysfunctions, and neurodevelopmental irregularities in real-time, promoting earlier and more effective clinical interventions. Together, AI and 4D ultrasound provide a dynamic approach to prenatal care, making the transition from reactive treatments to proactive and personalized interventions.[10]

Furthermore, early diagnosis increases the survival rate and decreases the complication rate.[11] However, the risks in healthcare are high to tolerate failures that could cause harm to patients. This means that, just like humans, AI models can be biased or make mistakes. Thus, AI combined with human intelligence in the diagnosis of CHDs, despite being beneficial, needs expandability to be implemented.[8]

Therefore, the AI system is capable of providing instant feedback through automatic structure recognition techniques, helping physicians to identify abnormalities more quickly and accurately, especially in complex and rare cases, where the assistance provided by AI would have the capacity to compensate for the deficiencies of inexperience. However, despite the great technological potential, more validation and clinical optimization measures are needed for it to be safely implemented.[4]

The collaborative role of AI and the physician in the diagnosis of fetal congenital heart defects

Fetal ultrasound is an operator-dependent exam, so an AI model, combined with human intervention, can be advantageous for improving fetal ultrasound screening for structural malformations, even though neither model achieves 100% accuracy. In this context, it is unlikely that AI can be used without the collaboration of a physician in conjunction. However, this technology, used to advise sonographers, culminates in significant improvement in diagnostic performance, according to recent studies.[5]

In this bias, a study that aimed to analyze the combined performance of AI and humans in the image diagnosis of fetal CHDs used ultrasounds with already proven heart anomalies and also images where the fetus had no anomalous heart condition. Such images were analyzed by two consultant fetal cardiologists, five non-physician sonographers, and three physicians with specialized postgraduate training in pediatric cardiology, with different years of experience in the area, who were compared with the AI developed. The results showed that more experienced physicians obtained superior results than artificial intelligence, but the clinical interaction and AI obtained satisfactory results, especially in relation to less experienced physicians. However, when AI presented incorrect results, it worsened the performance of the physicians, showing that this tool does not have 100% specificity and that the wrong results negatively impact the diagnosis.[5]

The use of AI in fetal cardiac evaluation reinforces that it is not necessary for only specialist physicians to perform fetal ultrasound exams and, thus, facilitates the diagnosis for other less experienced physicians. Currently, AI is not widely implemented clinically, being mostly used at an academic level for the prediction of preeclampsia and the detection of malformations and diagnosis of other conditions.[12]

Final Considerations

This work sought to synthesize the available evidence regarding the diagnosis of CHD with the aid of AI. In this context, the impact of the early identification of these pathologies on favorable clinical outcomes was analyzed, as well as the adversities that cause a significant rate of underdiagnosis with the use of fetal echocardiography.

When comparing the current screening model with the AI-assisted screening standard, it is evident that the applicability of artificial networks provides better results, with a reduction in the rate of underdiagnoses. However, intelligent computing models are still flawed and biased if used without human assistance.

The scope of congenital heart defect diagnosis with the aid of AI is still an area that lacks clearer clinical evidence. For a better elucidation of the theme, further studies are needed with the aim of better understanding the impact of AI in fetal cardiology.

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