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Diagnostic Accuracy of Machine Learning Models to Identify Congenital Heart Disease: A Meta-Analysis

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Background: With the dearth of trained care providers to diagnose congenital heart disease (CHD) and a high mortality burden, CHD lends itself to machine learning (ML) models that can improve the diagnostic accuracy of early screening tools, such as auscultation.

Methods: A comprehensive literature search in the PubMed, CINAHL, Wiley Cochrane Library, and Web of Science databases was performed. Studies that reported the diagnostic ability of ML models for the detection of CHD compared to the reference standard were included. Risk of bias assessment was performed using Quality Assessment of Diagnostic Accuracy Studies-2 tool. The sensitivity and specificity of the ML models for each outcome were used to generate the hierarchical summary ROC (HSROC) curves.

Results: Six included studies (enrolling 4,683 participants) used ML algorithms to diagnose CHD. Neural networks were used in seven studies and other ML algorithms (such as decision trees (DT) or logistic regression) in three studies. The ML models had a pooled sensitivity of 90.5% (95% CI: 86.1–93.8%) and specificity of 85.4% (95% CI: 77.9–90.8%). The corresponding HSROC curve demonstrated a better overall diagnostic ability of studies had a high risk of patient selection bias, sample sizes less than or equal to 1,000 (n ≤ 1,000), and fewer than 10 ML algorithms. The risk of bias was rated overall for the reference standard for six/ML.

Conclusions: ML models such as neural networks have the potential to diagnose CHD accurately without the need for an echocardiogram. Future studies should evaluate the diagnostic ability of these models when the heterogeneity of the CHD diagnoses included between the studies is a major limitation.

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