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Comparative Study on Early Detection of Congenital Heart Disease (CHD) Using Foetal Echocardiography

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A B S T R A C T

According to the World Health Organization, congenital heart defects are the cause of nearly one-third of all birth anomalies globally, in terms of neonatal morbidity and mortality. Due to climate change, children born to mothers suffer the cause from malnutrition and other health complications and are at greater risk of developing congenital heart defects as a result of variations in food availability and eating habits. Although early detection is critical, in general healthcare environments, several severe cardiac abnormalities are regularly overlooked by routine prenatal screening. This drawback, which results in poor neonatal outcomes and postponed detection, is mainly attributable to operator competence and differences in imaging quality.

Traditionally, foetal suffer foetal cardiac function has been defined using the Cardiovascular Profile score. The CVP score is of great value in diagnosing heart failure in utero. He suggested that an overall CVP score of ≤7 significantly predicts an unfavourable foetal outcome. In contrast, diagnostic systems powered by artificial intelligence have become a hopeful advancement since they can autonomously examine ultrasound images with a high degree of precision. However, a handful of methods that assess non-invasive maternal saliva biomarker analysis give fresh hope for detecting a certain CHD early.With accuracy levels above 90%, AI-driven image interpretation greatly lowers observer bias and increases detection consistency. This solution has potential to the potential improve the early and accurate identification of high-risk pregnancies when combined with straightforward, non-invasive biochemical screening techniques.

AI and biomarker-based methods work collectively to provide accurate prenatal cardiac screening. One of the main challenges to be overcome the potential before widespread adoption can take place is the interpretation of AI algorithms to ensure their ethical, equitable implementation. Also, addressing the environmental and nutritional effects of climate change may be essential to preventing foetal cardiac abnormalities in subsequent generations.

Keywords: Machine Learning, Foetal Echocardiography, Artificial Intelligence

Introduction

Congenital heart disease (CHD) is one of the most frequently occurring serious birth defects and a significant health problem globally. Pregnancy and Birth: About 8 to 9 of every 1,000 babies are born. These have a key endpoint that could include heart defects, which can be simple or complex and life-threatening combinations like the TOF and HLHS. Identifying these malformations before birth is exceedingly challenging.[1-4] It also assists parents in making more knowledgeable choices and ensures the best care for their baby immediately after delivery. This is critical in serious conditions such as CCHD, where delay can cause additional sickness and mortality.[3,4]

Although a specialised heart ultrasound of the baby (foetal echocardiography) can be quite accurate (over 90%) when performed by an expert, common use of it as a general foetal screening tool has led to varying and usually bad results. These large studies have shown that the overall detection rate is not higher than about a disappointing 68.5%, and it declines even further in routine low-risk pregnancy care where most screening occurs.[5] One This "diagnostic gap" is important because, of babies with the most serious heart defects, approximately 25% are overlooked at the time of screening and sent home, with many heartbreaking resulting deaths.[6]

One The failure is often due to the person performing the scan; they might not be skilled enough to get the right image of the heart, or they might miss a problem that's right there on the screen.1 This ongoing, system-wide failure highlights an urgent need for emerging computational methods that can help standardise the images and make diagnosis more reliable for everyone.[4,6]

This thorough analysis compares the disruptive innovations of AI-enhanced diagnosis and novel molecular screening using maternal metabolomics with conventional prognostic metrics, such as the Cardiovascular Profile (CVP) score. According to the review, AI is a vital translational tool for Precision Foetal Cardiology, helping to integrate multi-modal data streams and standardise diagnostic workflows.[7]

Table 1.Comparison between Traditional and AI-Based Methods for Detecting Fetal Heart Defects

Aspect	Traditional Methods	AI-Based Methods
Accuracy and Detection Rate	Detection rate remains low (around 30–40%) in non-specialist settings due to operator dependence and image quality variations.	Achieves over 90% accuracy by standardising image analysis and reducing observer bias.
Operator Dependency	Highly dependent on sonographer's skill, experience, and machine quality; results vary widely between clinics.	Minimises skill-based variation; algorithms provide consistent interpretation across users and machines.
Data Utilization	Relies mainly on 2D ultrasound images and manual scoring systems such as the Cardiovascular Profile (CVP).	Integrates multiple data streams including ultrasound images, Doppler data, and biochemical or molecular markers (e.g., saliva tests).
Speed and Efficiency	Interpretation and diagnosis can be time-consuming and require specialised fetal cardiologists.	Automated analysis provides real-time or near real-time feedback, speeding up screening and decision-making.
Predictive Capability	Effective mainly for known or advanced cases; limited in early prediction or subtle pattern recognition.	Excels at early detection and pattern recognition, identifying abnormalities invisible to the human eye.
Reproducibility	Results can vary due to subjective assessment and differing clinical environments.	Produces uniform and reproducible outcomes regardless of operator or location.
Accessibility	Limited in rural or resource-poor settings due to shortage of trained experts.	Can be extended via tele-echocardiography and remote AI-assisted interpretation, improving rural access.

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Ethical and Practical Challenges	Generally well-established and trusted but lacks scalability for mass screening.	Faces challenges with data privacy, explainability, and algorithmic bias, requiring strong ethical oversight.
Integration with Emerging Health Concerns	Rarely considers external or environmental factors affecting maternal health.	Capable of integrating maternal nutrition, environmental, and lifestyle data helpful in assessing climate change–related foetal risks.
Overall Outcome Potential	Provides valuable baseline assessment but often fails to catch critical defects early.	Represents a holistic, data-driven approach capable of supporting precision foetal cardiology and improving survival outcomes.

Literature Review

Standard Fetal Heart Check-up: Understanding the CVP Score

Because it allows for a sophisticated visualisation of the structure, function, and blood flow of the heart, foetal echocardiography continues to be the clinical cornerstone for the diagnosis of congenital heart disease.[2,7] Its intricacy and high level of operator dependence restrict its consistency in standard screening settings, despite the fact that it is extremely accurate in skilled hands. Foetal Congestive Heart Failure (CHF), which is closely linked to high perinatal mortality in cases of CHD, can be functionally assessed using the Cardiovascular Profile (CVP) score, a 10-point rating system.[4,8] One It incorporates five ultrasound markers: arterial umbilical Doppler, venous Doppler measurements, heart function (ventricular shortening fraction, SF, and regurgitation), heart size (heart-to-chest area ratio, HA/CA), and hydrops.[8]

Strong prognostic value is offered by the CVP score. The risk of death was statistically and significantly higher for foetuses with a final CVP score of ≤ 7 (87.5% mortality) than for those with a score of ≥ 8 (15.2% mortality). One Even after adjusting for covariates, this highly significant relationship results in a high odds ratio for death (OR 22.3; P=0.024) for scores ≤ 7. One On their own, severe cardiomegaly (HA/CA ratio > 0.5; OR 11.4) and hydrops (OR 12.4) are statistically significant markers of elevated mortality risk.[9]

The CVP score has a high specificity (up to 0.99 for mortality) and a notably low sensitivity (approximately 0.27 for mortality and 0.25 for poor Apgar score), despite its high value for risk stratification once CHD is known. One Therefore, the CVP score is mainly used to stratify risk in situations where CHD has already been suspected or confirmed, assisting in the decision making process regarding enhanced surveillance and delivery timing.[7,9]

Table 2.Structure and Prognostic Value of the Fetal Cardiovascular Profile (CVP) Score

Category (Max 2 Points)	Abnormal Finding (1 Point Deduction)	Severe Abnormality (2 Point Deduction / 0 Points)	Clinical Prognostic Significance
Hydrops	Ascites or pericardial/ pleural effusion	Skin edema (Hydrops Fetalis)	High specificity for perinatal mortality (OR 12.4).
Heart Size (HA/CA Ratio)	0.35 - 0.5	<0.2 or > 0.5 (Severe Cardiomegaly)	Severe cardiomegaly (> 0.5) strongly associated with mortality (OR 11.4).
Cardiac Function (SF/ Doppler)	Holosystolic TR or LV/RV SFs < 0.28	Holosystolic MR or TR dp/dt < 400 , Monophasic filling	Direct metric of myocardial dysfunction/CHF.
Arterial Umbilical Doppler	Absent End-Diastolic Velocity (AEDV)	Reversed End Diastolic Velocity (REDV)	Reflects compromised placental perfusion/increased afterload.
Venous Doppler	Ductus Venosus (DV) atrial reversal	Umbilical Vein (UV) pulsations	Sign of elevated central venous pressure.

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Artificial Intelligence in Fetal Cardiology: Standardizing and Diagnosing

The fundamental issue of operator variability, which leads to the diagnostic crisis in CHD screening, can be resolved with the quick uptake of artificial intelligence (AI) and deep learning (DL). Complex image processing and pattern recognition are two areas in which DL, and more especially Convolutional Neural Networks (CNNs), shine.[3,9]

AI for Image Quality

Right now, the most urgent and helpful job for AI is to handle the basic technical parts of getting a good image, making the whole process more standard and reliable. For instance, new AI models can watch an ultrasound video feed and automatically find and save the most important views of a baby's heart, like the four-chamber view or the pathways where blood flows out.[10]

This is a game-changer because it lets the human expert focus their attention on spotting any problems, instead of struggling to get the perfect picture. It also helps make up for differences in skill between the people performing the scans.[11]

AI algorithms also play a key role in quality control, making sure that only sharp, clear images are used to make a diagnosis. But AI is great at more than just picking pictures; it also excels at measuring things.[8,11] It can accurately map out the heart's complex 3D structures and automatically calculate key health indicators (like heart chamber volumes or how much blood is being pumped). This is vital for tracking a baby's condition over time.[12]

Finally, even emerging computational methods like ChatGPT are showing they can help by taking a few measurements and brief clinical notes and then generating a complete, accurate report. This saves a huge amount of time on paperwork and reduces the differences in how various doctors might describe the same results.

AI for Diagnosis

It turns out that when you train an AI on a huge library of past medical cases, it gets incredibly good at spotting heart defects—so good, in fact, that it's now performing at the same level as a human expert.[9,12] One It's remarkably sharp at telling a healthy heart from one with a complex problem, correctly identifying the issue more than 90% of the time and hitting performance scores (AUROC values) as high as 0.99, which is almost perfect.[13]

Because it's so reliable, this kind of AI could be the key to solving our current problem of missed diagnoses. For a specific, very serious condition like Hypoplastic Left Heart Syndrome (HLHS), the models are right around 94.3% of the time. And this isn't just for prenatal screening, either. The same technology is being used after birth to help predict the chances of success for incredibly high-risk surgeries, like the Norwood procedure, with an impressive accuracy score of about 0.95.[14]

Precision Fetal Cardiology: Molecular Diagnostics

A huge challenge in medicine is that most babies with heart defects are born to mothers with no known risk factors. This is where a potential game-changer comes in: using AI to screen for these conditions by analysing the tiny molecules in a mother's saliva. This new approach has shown incredible promise for finding a serious group of heart defects known as cyanotic CHD (CCHD). When tested, it proved to be highly accurate, correctly spotting the condition up to 92.5% of the time while also correctly giving the all-clear to healthy pregnancies up to 91.0% of the time[14]. The overall performance scores (the AUC) are consistently strong, ranging from about 0.82 to 0.85.

The best part is that a saliva test is easy, painless, and perfect for screening a large number of people. It also gives doctors a completely new source of information that doesn't rely on getting a perfect ultrasound picture.[15]

When scientists looked deeper into the saliva samples, they discovered something fascinating. The data pointed to a significant disruption in how the body was processing certain fats—specifically arachidonic acid and alpha-linolenic acid. This suggests that a system-wide problem with fat metabolism is linked to these heart defects in the baby. One This is a massive shift in thinking. It means we could move from just identifying the physical damage the disease causes to detecting the hidden biological process that sets it all in motion. This very idea—targeting the root molecular cause—is creating the foundation for a whole new field: Precision foetal Cardiology.[15]

Discussion

Bridging the Diagnostic Gap and Synthesising Data

Prenatal cardiac screening could be revolutionised through the practical and potent potential of artificial intelligence (AI). A significant percentage of congenital heart defects (CHDs) go undetected until birth due to the startlingly low detection rates that are currently in place in typical clinical settings, which frequently hover around 36%. AI-based systems, on the other hand, have shown accuracy levels above 90%, mainly due to the fact that they remove discrepancies brought about by operator experience, equipment unreliability, and subjective image interpretation.[14]

The ability of AI to synthesise various data streams is what gives it its true value, not just its accuracy. Traditional instruments like the Cardiovascular Profile (CVP) score are still very useful for assessing foetal risk and tracking the

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course of the disease, but Deep Learning (DL) algorithms are excellent at spotting subtle structural and functional abnormalities during early screening.[10,14] When combined, these two modalities help close the sensitivity–specificity gap that frequently restricts traditional methods.

Precision foetal cardiology needs a system of diagnostics in its entirety. This framework should integrate real-time ultrasound images, AI analysis, CVP scoring, and molecular biomarkers (including saliva-based tests) into a single platform to make decisions. By creating links between physiological, anatomical and biochemical indicators, clinicians would cause a transition between population-based and customised foetal risk prognosis. This model would allow truly personalised prenatal care plans on top of raising diagnostic accuracy.[14,15]

In addition, the effect of climate change on maternal health is one of the aspects that are usually ignored. Food systems and nutritional patterns are already being affected by environmental disturbances all over the world. Alterations in food supply and quality may cause maternal deficiencies in micronutrients, which may consequently expose the foetus to the risk of congenital heart defects.[15] With the inclusion of environmental and nutritional variables into the AI-based prediction models, an essential preventive dimension would be introduced, and the technology would consider the emerging health trends in the world.[15]

Challenges to Clinical Translation

The path to integrating AI-based foetal cardiac diagnostics into routine clinical practice is still difficult, despite its potential. In addition to technology, there are logistical, moral, and social obstacles.

The Challenge of Limited and Inconsistent Data

One of the biggest problems is that we just don't have enough resources or standard, well-labelled datasets. Data from different hospitals is all over the place; they use different gear, label things differently, and the image quality varies by area. So, a model built in one place probably won't work well somewhere else. And since CHDs are so diverse and pretty rare, these small, scattered datasets just can't cover the full picture.[15]

For better results, we really need to create central, high-quality registries for foetal CHD cases and get everyone to share data. Making terminology, imaging, and labelling standards the same everywhere would also be a huge help for reliability. There's a practical fix, too, using things like federated learning. It lets different organisations train shared AI models together, but without ever sending private patient info. It may be a great way to balance being inclusive and innovative while still protecting patient confidentiality.[11,15]

Challenges of Trust and Ethics in AI Deployment

The opaqueness of AI decision-making procedures is a significant obstacle to clinical adoption. Many algorithms function as model interpretability challenges, producing results without providing explicit justifications for their methodology. This makes it difficult for clinicians to trust one another. Doctors need simplified and trustworthy systems before implementing AI-generated reports into critical health decisions.[15] As a result, Explainable AI (XAI)—technologies that make model reasoning traceable and intelligible—is receiving more attention.

Attention must also be paid to equity and ethical issues. Algorithms run the risk of introducing biases that lower diagnostic accuracy for under-represented populations if training datasets are not diverse in terms of demographics. Furthermore, by concentrating advanced care in high-income or urban areas, the expense and technical demands of AI-based diagnostics may worsen healthcare disparities.[6,15]

A hybrid approach that combines AI and tele-echocardiography could expand expert-level screening to underprivileged areas in order to close these gaps. AI-assisted remote interpretation by experts would lessen regional disparities and democratise access to prenatal cardiac care.[8]

Conclusion

By offering workable answers to the significant diagnostic gaps present in the current screening techniques, artificial intelligence (AI) presents a game-changing potential for enhancing the diagnosis and treatment of foetal congenital heart disease [15]. AI serves as the foundation for Precision Foetal Cardiology by improving prenatal imaging sensitivity (from as low as 36.1% to greater than 90%) through automated quality control and view classification, as well as by incorporating functional prognostic metrics like the CVP score. Additionally, by detecting molecular vulnerabilities connected to the pathophysiology of CCHD, AI-driven analysis of maternal metabolomics adds a non-invasive layer of screening. Algorithmic transparency (XAI) must be prioritised to build clinician trust, data standardisation must be accelerated to ensure model generalisation, and ethical and regulatory frameworks must be flexible to support continuously learning models and ensure equitable access to these life-saving advancements.[7,15] These issues must be resolved in order for the clinical integration of these technologies to be successful.

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