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MODERN DIAGNOSTIC METHODS FOR EARLY DETECTION OF CONGENITAL HEART DEFECTS IN CHILDREN

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Abstract: Background: Congenital heart defects (CHDs) encompass a spectrum of anatomical abnormalities, including lesions requiring intervention immediately after birth. Objective: To compare contemporary prenatal, neonatal, and pediatric diagnostic approaches and propose a staged referral algorithm suitable for resource-variable care settings. Methods: A narrative, structured synthesis of international screening studies, professional guidance, and Uzbekistan-based publications was undertaken. Results: Fetal cardiac imaging detects many complex lesions before delivery; postnatal right-hand/foot pulse oximetry identifies hypoxemia associated with some critical CHDs, whereas echocardiography establishes anatomy and hemodynamics. Evidence indicates excellent screening specificity but incomplete sensitivity, necessitating continued clinical examination and follow-up. Conclusion: Combining risk-based prenatal referral, newborn clinical examination, pulse oximetry, timely echocardiography, and a clear regional referral network is more defensible than reliance on any single test.

Keywords: congenital heart defect; prenatal ultrasound; fetal echocardiography; pulse oximetry; newborn screening; pediatric echocardiography; early diagnosis.

Introduction

Congenital heart defects (CHDs) are structural abnormalities of the heart or great vessels present at birth. Their presentations range from small septal defects to critical lesions with duct-dependent systemic or pulmonary circulation. The diagnostic window matters: a newborn may appear stable during the first hours of life, then deteriorate as the ductus arteriosus closes. Accordingly, early diagnosis is not simply a matter of identifying a cardiac murmur. It requires recognition of risk factors, prenatal anatomical assessment, postnatal physiologic screening and access to confirmatory cardiovascular imaging (1, 2).

The clinical burden is particularly important for regions where specialist echocardiography, transport and surgical resources are unevenly distributed. Evidence from Uzbekistan describes differences in diagnostic pathways across district, regional and republican levels, while regional work in Andijan documents a substantial group of children evaluated for CHDs (11, 13). These observations motivate a tiered approach that specifies which test is appropriate at which point, and which finding should trigger urgent referral.

This study aimed to describe the comparative roles, advantages and limitations of modern diagnostic methods and to propose an evidence-informed pathway for detecting CHDs prenatally and throughout early childhood. The paper is a literature-based analysis, not a newly conducted clinical trial.

Literature review

International research established that pulse oximetry supplements, but does not replace, newborn examination. A meta-analysis by Thangaratinam and colleagues, involving 229,421 infants in 13 studies, reported pooled sensitivity of 76.5% and specificity of 99.9% for critical CHD screening (3). Ewer and colleagues evaluated the performance of neonatal pulse oximetry in a large prospective study, supporting its use alongside conventional assessment (4). However, diagnostic sensitivity varies with lesion type, timing and screening population; a single-center report demonstrated that even highly specific protocols can miss important defects (5).

Contemporary CDC guidance explicitly requires examination of the right hand and foot and interpretation of saturation and inter-limb difference using a defined algorithm. Screening after approximately 24 hours is preferred when feasible; a passing result does not exclude all CHDs (2). Prenatal four-chamber and outflow-tract views, together with specialist fetal echocardiography for selected pregnancies, improve identification before birth and enable delivery planning. Detailed pediatric transthoracic echocardiography remains the principal confirmatory examination after birth, complemented by ECG, radiography and, when necessary, CT or MRI (6–10).

Uzbekistan-based publications add health-system context. Islamov and coauthors compared echocardiographic diagnosis at different levels of pediatric care (11). Akhmedova and Sotvoldieva reviewed epidemiology, screening and risk factors (12), while an Andijan regional cohort described the distribution of surgically managed lesions (13). Rozikhodjaeva and Nazirkhujaev emphasized the importance of locally appropriate pediatric echocardiographic reference parameters (14); other local authors discuss 3D imaging, speckle tracking and age-specific Z-scores as supportive tools (15–17). These works are informative but differ in scope and design, so their findings should not be combined as if they formed one standardized national screening cohort.

Materials and methods

A narrative analytical design was used. Sources included peer-reviewed studies, public clinical guidance and locally authored work published or accessible by October 2026. The conceptual comparison covered (a) prenatal detection, (b) initial newborn examination, (c) pulse-oximetry screening, (d) definitive echocardiography and (e) advanced cross-sectional imaging. Evidence was synthesized by clinical purpose, timing, principal strengths and important limitations. No patients were enrolled, no unpublished measurements were created and no statistical testing was performed on original clinical observations.

The bar diagram reproduces two published pooled test-performance estimates and the lower boundaries of their reported 95% confidence intervals from reference (3). It is not an original clinical dataset, does not compare screening devices, and does not indicate the accuracy of fetal ultrasound or echocardiography. This restricted presentation avoids implying equivalence among tests used for different clinical purposes.

Results

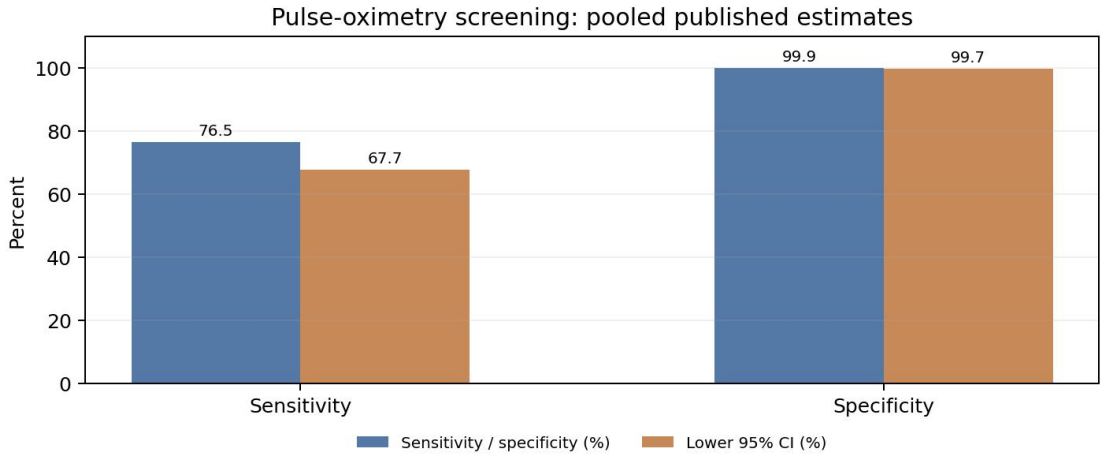
The synthesis yielded a five-stage diagnostic pathway. During pregnancy, routine obstetric ultrasound should examine cardiac situs, chambers and major outflow tracts; suspected anomalies or relevant maternal/fetal risk factors require specialist fetal echocardiography. At birth, clinical assessment includes color, respiratory effort, feeding tolerance, pulses, perfusion, auscultation and

family/prenatal history. For asymptomatic infants without an established diagnosis, standardized preductal and postductal pulse oximetry can help detect occult hypoxemia. Abnormal results or concerning symptoms demand timely medical assessment and usually echocardiography. After stabilization, pediatric cardiology teams determine anatomical diagnosis, urgency and further imaging needs (2, 6, 11).

Table 1. Diagnostic methods and clinical roles

Method / period	Principal value	Important limitation
Obstetric ultrasound / antenatal	Screening of chamber and outflow-tract anatomy	Operator and gestational-age dependent
Fetal echocardiography / antenatal	Detailed lesion mapping; delivery planning	Not all lesions evident prenatally
Clinical examination / birth onward	Finds cyanosis, pulse deficits and heart failure	Some critical lesions initially silent
Pulse oximetry / newborn	Detects hypoxemia in some critical CHDs	Can miss nonhypoxemic lesions
Transthoracic echo / suspected CHD	Defines anatomy, shunt and hemodynamics	Depends on expertise and acoustic windows
CT/MRI / selected complex CHD	Maps extracardiac vessels and 3D anatomy	Access, sedation or radiation considerations

Figure 1. Published pooled pulse-oximetry screening estimates (3)



The figure highlights an asymmetric screening profile: high specificity limits false alerts, but imperfect sensitivity means a normal reading cannot rule out serious disease. The 95% interval bounds relate to the pooled meta-analysis, not to an Uzbekistan-specific screening program. Results should not be extrapolated to all CHD categories or to ultrasound performance (3).

Table 2. Suggested actions after initial screening and examination

Finding	Clinical response
Known prenatal CHD or shock/cyanosis	Immediate neonatal-cardiology assessment; stabilize and obtain urgent echo.
Failed pulse-oximetry screen	Evaluate hypoxemia promptly; consider

	echocardiography and alternative causes.
Normal saturation but weak femoral pulses or murmur	Do not dismiss CHD; perform focused exam and refer for echo as indicated.
Normal screen and examination	Continue routine care and communicate warning signs to caregivers.

A practical referral model should document who interprets screening, who can perform echocardiography, how images are shared and how transport is arranged. Digital archiving and tele-echocardiography may improve communication between district facilities and specialized centers, but they require staff training, privacy safeguards and local quality control (11, 14, 16).

Discussion

Prenatal and newborn testing are complementary rather than interchangeable. Prenatal imaging can identify many complex structural anomalies and make delivery at a cardiac center possible; however, changes in circulation after birth may reveal previously unrecognized obstruction or shunting. Therefore, neonatal examination and physiologic screening remain important even after apparently reassuring antenatal scans. Conversely, once a fetus has a clear cardiac diagnosis, the purpose of postnatal assessment is confirmation and clinical stabilization, not repeating routine screening as a substitute for an urgent cardiology plan (2, 6, 8).

The principal value of pulse oximetry is its simplicity and ability to detect hypoxemia before obvious clinical deterioration in some infants. Yet the test identifies physiological consequences, not cardiac morphology. Left-sided obstructive lesions, including some coarctations, may pass screening. An algorithm that equates normal oxygen saturation with a normal cardiovascular system is therefore unsafe. The strongest approach combines observation, pulse examination, saturation and selective echocardiography (2–5).

Echocardiography is central to definitive diagnosis because it delineates septal defects, valve malformations, ventricular function and blood-flow patterns at the bedside. Doppler imaging adds pressure-gradient and flow information, while advanced 3D methods and speckle tracking may aid selected assessments. These techniques do not eliminate the need for conventional structural evaluation; interpretation requires pediatric-specific measurement references and attention to body size, age and hemodynamic conditions (7, 14–16). Cross-sectional angiography is reserved for anatomically complex cases in which a complete depiction of pulmonary veins, aorta or collateral vessels is needed.

The main limitation of this synthesis is heterogeneity among the included publications. Screening programs differ in population, timing and definitions of a positive test, while Uzbekistan studies include reviews, tertiary-center cohorts and technology discussions. Consequently, this paper does not estimate a national detection rate or prove the effectiveness of any particular intervention. A future prospective multicenter project should record denominator data, lesion subtype, time to diagnosis, follow-up of negative screens, and outcomes after referral (11–13).

Conclusion

Early detection of congenital heart defects in children requires a coordinated sequence of prenatal ultrasound, risk-based fetal echocardiography, careful neonatal examination, standardized pulse-oximetry screening and prompt transthoracic echocardiography when indicated. Screening performance must be interpreted with lesion-specific limitations; negative pulse oximetry does not

exclude CHD. For Uzbekistan, consistent referral pathways, access to pediatric specialists, locally validated echocardiographic reference values and data-quality monitoring are practical priorities. The proposed approach is evidence-informed and should be evaluated in regional implementation studies before claims of improved outcomes are made.

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