PRENATAL DIAGNOSIS OF CONGENITAL HEART DEFECTS: DETECTION OPPORTUNITIES AND INITIAL ACTION PLAN

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Abstract: *This article examines modern approaches to prenatal (antenatal) diagnosis of congenital heart defects (CHD) in the fetus. It emphasizes the key role of prenatal screening in the early detection of heart abnormalities, which directly impacts postnatal prognosis and survival. The article analyzes the diagnostic capabilities of different ultrasound techniques (first and second trimester screening, fetal echocardiography), as well as the potential of additional methods (genetic testing, magnetic resonance imaging). A structured initial action plan for healthcare providers and families upon detection of a suspected heart defect is presented. This includes stages of further verification, genetic counseling, multidisciplinary consultation (perinatologist, pediatric cardiologist, cardiac surgeon), and planning for delivery in a specialized perinatal center. Early diagnosis and a clear action plan can optimize perinatal management, improve surgical outcomes, and provide crucial psychological preparation for parents.*

Keywords: *Congenital heart defects, prenatal diagnosis, fetal echocardiography, antenatal screening, ultrasound of the fetus, perinatal management, genetic counseling, perinatal cardiology, action plan, multidisciplinary approach.*

Introduction

Congenital heart defects remain the most common group of severe congenital malformations, affecting approximately 6-10 per 1000 live births worldwide and

representing a leading cause of infant mortality [Hoffman, Kaplan, 2002, p. 120]. The development of modern perinatal diagnostics has fundamentally changed the approach to managing these conditions. While postnatal diagnosis often leads to emergency situations and delayed surgical care, prenatal detection offers the opportunity to plan, prepare, and optimize all stages of care for the child. The primary goal of prenatal screening for congenital heart defects is not just to detect an abnormality, but to initiate a cascade of specialized measures aimed at ensuring the best possible outcome for the newborn. Early diagnosis allows for the determination of the need and timing for delivery in a specialized facility with a pediatric cardiac surgery unit, prevents the catastrophic consequences of critical congenital heart defects after ductus arteriosus closure, and enables the family to receive comprehensive information and psychological support. This article aims to analyze the current possibilities of prenatal detection of congenital heart defects and to propose a structured, step-by-step action plan for use after detection.

Literature Review

The history of fetal heart diagnostics began with the establishment of M-mode echocardiography. A breakthrough was the development and implementation of high-resolution two-dimensional ultrasound, color and pulsed Doppler mapping, which formed the basis of modern fetal echocardiography. Landmark studies, such as those by [Allan, 2000], proved that systematic examination of the fetal heart during a routine second-trimester scan can identify up to 60-70% of major congenital heart defects. However, sensitivity varies significantly depending on the examiner's qualification, the protocol used, and patient-specific factors (maternal obesity, fetal position). The importance of first-trimester screening is highlighted in the works of [Carvalho et al., 2013], which demonstrated the possibility of detecting certain major defects as early as 11-14 weeks by assessing the four-chamber view and outflow tracts. The evolution of screening protocols towards extended basic cardiac examination has been a subject of discussion in consensus documents from international societies such as ISUOG [International Society of Ultrasound in

Obstetrics and Gynecology, 2013]. The role of 3D/4D ultrasound and spatiotemporal image correlation (STIC) remains complementary but can be valuable for clarifying complex anatomy and for educational purposes. Beyond imaging, the literature emphasizes the tight link between congenital heart defects and genetic syndromes. Studies show that up to 25-30% of fetuses with congenital heart defects have an associated chromosomal abnormality or monogenic syndrome [Pierpont et al., 2018, p. 2243]. This underscores the critical necessity of integrating genetic counseling and testing (karyotyping, chromosomal microarray analysis) into the diagnostic algorithm. Post-diagnostic management is thoroughly covered in the works of [Donofrio et al., 2014], who developed guidelines for the delivery planning and perinatal care of fetuses with congenital heart defects, highlighting the need for a "delivery planning conference" involving all key specialists.

Discussion

The prenatal detection of congenital heart defects is a multi-stage process that begins with population-based screening and ends with highly specialized diagnostics and delivery planning.

1. **Diagnostic Modalities and Their Capabilities.**

Routine Obstetric Ultrasound Screening: This is the primary screening tool. The first-trimester scan (11-14 weeks) can identify gross abnormalities and markers like increased nuchal translucency, which is associated with a higher risk of congenital heart defects and aneuploidies. The mandatory second-trimester anatomical survey (18-22 weeks) must include a detailed cardiac examination. The minimum standard is the assessment of the four-chamber view and visualization of the outflow tracts. The sensitivity of screening performed by trained sonographers using a standardized protocol can reach 85-90% for major congenital heart defects [International Society of Ultrasound in Obstetrics and Gynecology, 2013, p. 12].

Fetal Echocardiography This is a targeted, comprehensive examination performed by a specialist (fetal cardiologist/maternal-fetal medicine specialist with appropriate training). Indications include: suspicious findings on a routine scan,

family history of congenital heart defects, maternal diabetes, exposure to teratogens, fetal extracardiac anomalies, or arrhythmia. Fetal echocardiography assesses cardiac anatomy in detail, function, rhythm, and hemodynamics using 2D, color Doppler, and spectral Doppler.

Genetic Diagnostics - Upon detection of a congenital heart defect, invasive prenatal diagnosis (amniocentesis, chorionic villus sampling) with karyotyping and chromosomal microarray analysis is strongly recommended. This allows for the identification of associated syndromes (e.g., Down, DiGeorge, Turner), which significantly impacts prognosis and parental counseling.

Fetal Cardiac MRI - A promising but still secondary method, used primarily for complex defects or when ultrasound visualization is limited. It can provide excellent anatomical detail, especially for extracardiac vascular anatomy.

2. **Structured Initial Action Plan.** Upon a suspected or confirmed diagnosis of a congenital heart defect, a clear algorithm must be activated:

Step 1: Verification and Detailed Diagnosis. The pregnant woman is referred for a confirmatory expert fetal echocardiography. The exact anatomical diagnosis, its severity, and potential associated findings are established.

Step 2: Multidisciplinary Consultation and Counseling. A council is formed, ideally including a perinatologist/maternal-fetal medicine specialist, a pediatric cardiologist, a pediatric cardiac surgeon, a neonatologist, and a geneticist. The council discusses:

✚ Accurate diagnosis and prognosis (potential for biventricular vs. univentricular repair).
✚ Association with genetic syndromes.
✚ Planned postnatal management (need for prostaglandin infusion, timing of surgery).
✚ Overall prognosis for survival and long-term quality of life.

Step 3: Genetic Counseling and Testing. Parents are provided with detailed information about the genetic risks and offered invasive testing. The results are integrated into the overall prognosis.

Step 4: Development of a Perinatal Management Plan. This is the key practical outcome. The plan includes:

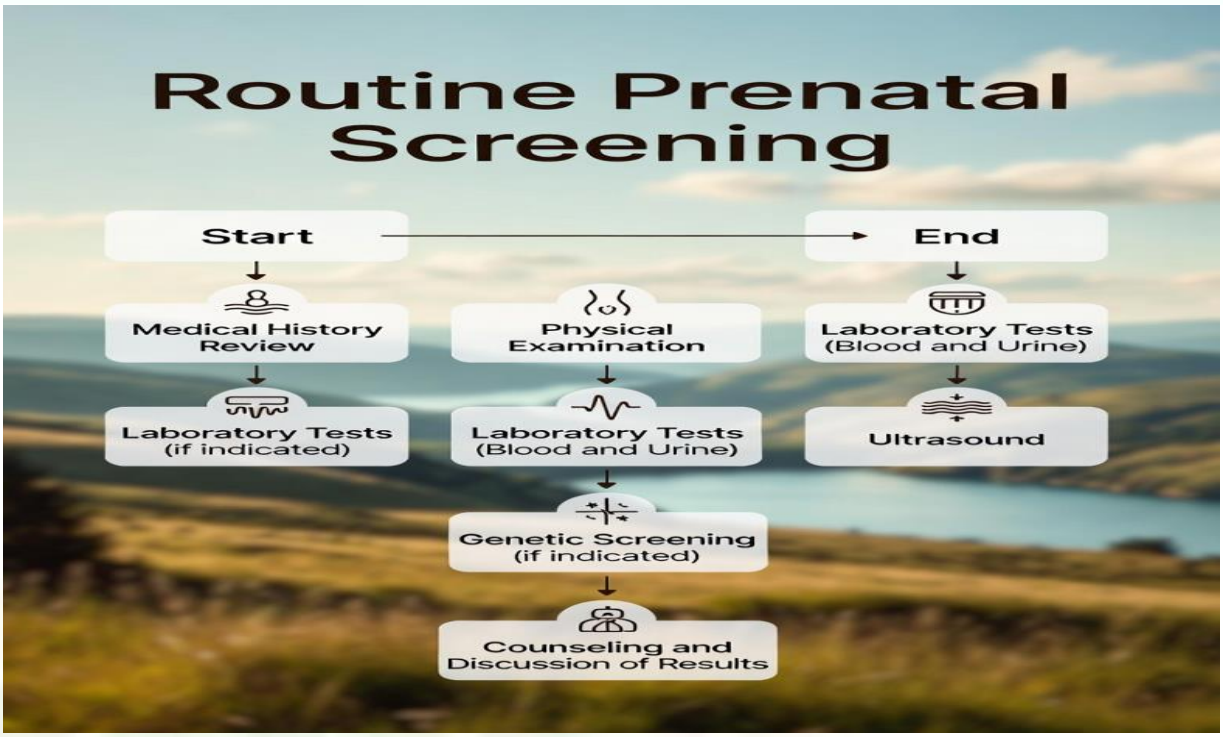
- Choice of Place of Delivery: Mandatory delivery in a Level III-IV perinatal center with an adjacent pediatric cardiac intensive care unit and surgery department.
- Timing and Mode of Delivery: In most cases, vaginal delivery at term is recommended. Caesarean section is reserved for obstetric indications or specific cardiac conditions (e.g., large fetal teratoma).
- Plan for the First Minutes/Hours of Life: Clear instructions are prepared for the neonatal resuscitation team regarding the need for immediate administration of prostaglandin E1 (for duct-dependent lesions) or other specific interventions.

Step 5: Psychological Support for the Family. From the moment of diagnosis, parents should have access to psychological counseling. Honest, compassionate, and consistent information from the medical team is the foundation for adaptation.

Results

The analysis of current prenatal diagnostic capabilities and management strategies allows for the formulation of key results. The successful implementation of an early detection and action system leads to measurable positive outcomes. The main effect is the transformation of a potentially catastrophic neonatal emergency into a planned, managed perinatal process. The data presented in the diagram below summarizes the key stages and outcomes of implementing a structured prenatal action plan for congenital heart defects.

Diagram: Algorithm and Outcomes of Prenatal Diagnosis and Management of Congenital Heart Defects



Conclusion

Prenatal diagnosis of congenital heart defects is a powerful tool of modern perinatology that has shifted the paradigm of care from emergency response to planned management. The capabilities of ultrasound screening, supplemented by expert fetal echocardiography and genetic testing, make it possible to detect the vast majority of severe heart defects in utero. However, detection alone is not enough. Its maximum effectiveness is realized only within the framework of a clear, structured action plan. This plan must include mandatory multidisciplinary consultation, comprehensive parental counseling, genetic diagnosis, and, most importantly, detailed planning for delivery and the immediate postnatal period in a specialized center. Such an integrated approach significantly reduces neonatal mortality and morbidity, improves the conditions for subsequent surgical correction, and provides invaluable psychological preparation for the family. Therefore, the development and universal implementation of standardized protocols for prenatal detection and management of congenital heart defects should be considered a priority in modern obstetric and pediatric cardiac care.

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