








Prenatal Diagnosis of Fetal Cardiac Anomalies: Diagnostic Concordance with Postnatal Echocardiography and Perinatal Outcomes

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Abstract

Objective: This study aimed to evaluate the concordance between prenatal and postnatal diagnoses in pregnant women with detected fetal cardiac anomalies and to present the perinatal outcomes of these pregnancies.

Methods: A total of 9,755 fetal echocardiographic examinations performed at our hospital's perinatology clinic between January 2019 and May 2025 were retrospectively analyzed. Ninety-two fetuses with structural cardiac anomalies identified during these examinations were included in the study. Maternal demographic characteristics, prenatal ultrasound and echocardiographic findings, the presence of accompanying extracardiac or genetic anomalies, pregnancy outcomes, and postnatal echocardiography results were examined. The concordance between prenatal and postnatal cardiac diagnoses in live-born infants was analyzed.

Results: The mean gestational age at diagnosis was 23.07±4.15 weeks. Septal defects were the most common cardiac anomalies (45.7%), followed by conotruncal defects (18.5%) and left-sided obstructive lesions (15.2%). Due to severe cardiac and/or associated extracardiac or genetic anomalies, pregnancy was terminated in 10 cases (10.9%). Prenatal and postnatal cardiac diagnoses were consistent in 72 of the 82 live births (87.8%), whereas inconsistencies were detected in 10 cases (12.2%). These inconsistencies were most commonly associated with small or muscular ventricular septal defects. Eleven deaths were observed in the postnatal period.

Conclusion: Prenatal diagnosis of fetal cardiac anomalies is important for the early detection of associated structural and genetic anomalies. Accurate diagnosis supports informed decision-making regarding the continuation or termination of pregnancy. Furthermore, early diagnosis of severe cardiac defects allows delivery to be planned in well-equipped centers and enables rapid postnatal intervention, which may positively influence neonatal outcomes. These findings emphasize the importance of prenatal echocardiography and a multidisciplinary approach in the perinatal management of congenital heart disease.

Keywords: Congenital; echocardiography; fetus; heart defects; infant; newborn; pregnancy outcome; prenatal diagnosis.

Fetal Kardiyak Anomalilerin Prenatal Tanısı: Postnatal Ekokardiyografi ile Tanısal Uyum ve Perinatal Sonuçlar

Özet

Amaç: Bu çalışmanın amacı, konjenital kardiyak anomalisi olan fetüslerde prenatal ve postnatal ekokardiyografik bulgular arasındaki tanısal uyumu değerlendirmek ve perinatal sonuçları incelemektir.

Yöntem: Ocak 2019–Mayıs 2025 tarihleri arasında perinatoloji polikliniğimizde gerçekleştirilen 9.755 fetal ekokardiyografi incelemesinin sonuçları retrospektif olarak değerlendirildi. Yapısal kardiyak anomalisi saptanan 92 fetüs çalışmaya dâhil edildi. Anneye ait demografik özellikler, prenatal ultrason bulguları, eşlik eden

Cite This Article: Yılmaz A, Genç FA, Kurt D, Dizdaroğulları GE, Bağlar İ, Kahyaoğlu B, Çeçen Y. Prenatal Diagnosis of Fetal Cardiac Anomalies: Diagnostic Concordance with Postnatal Echocardiography and Perinatal Outcomes. Koşuyolu Heart J 0000;00(0):1–7

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Submitted: January 16, 2026

Revised: March 13, 2026

Accepted: April 10, 2026

Available Online: June 02, 2026



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ekstrakardiyak ve genetik anomaliler, gebelik sonuçları ve postnatal ekokardiyografik bulgular incelendi. Canlı doğan bebeklerde prenatal ve postnatal kardiyak tanılar arasındaki tanısal uyum değerlendirildi.

Bulgular: Tanı anındaki ortalama gebelik yaşı $23,07 \pm 4,15$ hafta idi. En sık saptanan kardiyak anomaliler septal defektlerdi (%45,7); bunu konotrunkal defektler (%18,5) ve sol taraflı obstrüktif lezyonlar (%15,2) izledi. Şiddetli kardiyak ve/veya eşlik eden ekstrakardiyak ya da genetik anomaliler nedeniyle 10 olguda (%10,9) gebelik sonlandırıldı. Canlı doğan 82 bebekte prenatal ve postnatal kardiyak tanılar 72 olguda (%87,8) uyumlu iken, 10 olguda (%12,2) uyumsuzluk saptandı; bu uyumsuzluklar en sık küçük veya musküler ventriküler septal defektlerle ilişkiliydi. Postnatal dönemde 11 ölüm kaydedildi.

Sonuç: Fetal kardiyak anomalilerin prenatal tanısı, eşlik eden yapısal ve genetik anomalilerin erken saptanmasına olanak sağlamakta ve gebeliğin devamı ya da sonlandırılması konusunda bilinçli karar verilmesini desteklemektedir. Ayrıca şiddetli kardiyak defektlerin erken tanınması, doğumun ileri merkezlerde planlanmasına ve postnatal dönemde hızlı müdahaleye imkân tanılarak neonatal sonuçları olumlu yönde etkileyebilir. Bu bulgular, konjenital kalp hastalıklarının perinatal yönetiminde prenatal ekokardiyografinin ve multidisipliner yaklaşımın önemini vurgulamaktadır.

Anahtar sözcükler: Konjenital; ekokardiyografi; fetüs; kalp defektleri; infant; yenidoğan; gebelik sonucu; prenatal tanı.

Introduction

Fetal cardiac anomalies are the most common congenital structural malformations and account for a large proportion of birth defects.^[1,2] The reported incidence ranges from 0.8% to 1.2%, corresponding to approximately 8–12 cases per 1,000 live births.^[1,2] Several factors have been shown to increase the risk of fetal cardiac anomalies. These include perinatal infections, obesity, teratogenic exposures, pregestational diabetes, a positive family history of congenital heart disease, and chronic maternal diseases.^[3–5]

Prenatal diagnosis of fetal cardiac anomalies is critical, particularly in severe and life-threatening cases. Conditions such as hypoplastic left heart syndrome, transposition of the great arteries, and tetralogy of Fallot may require medical or surgical intervention soon after birth. In such cases, prenatal diagnosis allows delivery to be planned in tertiary centers with a neonatal intensive care unit, pediatric cardiologists, and cardiac surgery teams.^[6,7]

Even in milder forms of cardiac anomalies, prenatal diagnosis is valuable for evaluating the presence of syndromic or chromosomal associations, guiding perinatal care, and planning postnatal follow-up. Nonetheless, the effect of prenatal diagnosis on long-term survival remains a matter of debate.^[8,9]

The aim of this study was to assess perinatal outcomes and the diagnostic accuracy of prenatal detection of fetal cardiac anomalies at a tertiary healthcare center. The findings are expected to contribute to the literature by providing insight into the effectiveness of prenatal diagnosis in clinical practice and by guiding future perinatal management strategies.

Materials and Methods

Study Design and Setting

This retrospective study analyzed pregnancies diagnosed with fetal cardiac anomalies at our perinatology outpatient clinic between January 2019 and May 2025. Ethical approval was obtained from the Koşuyolu High Specialization Training and Research Hospital Scientific Research Board (Approval Number: 2025/09/1147, Date: 03.06.2025), and all procedures were conducted in accordance with the principles of the Declaration of Helsinki.

Patient Selection and Exclusion Criteria

The study cohort consisted of pregnant women diagnosed with fetal cardiac anomalies through routine prenatal ultrasound and fetal echocardiography who were followed at our perinatology outpatient clinic during the study period. Medical records from 9,755 fetal echocardiographic examinations were retrospectively reviewed, and 92 fetuses with structural cardiac anomalies were included in the study. Both singleton and multiple pregnancies were included in the analysis.

Patients were included if a structural cardiac anomaly was identified prenatally and sufficient prenatal and postnatal data were available to allow comparison of the diagnoses. Cases with incomplete medical records or those lost to follow-up before delivery were excluded.

Only fetuses with adequate imaging quality and complete documentation permitting reliable prenatal–postnatal diagnostic correlation were included in the final analysis. Isolated fetal arrhythmias or conduction abnormalities without associated structural cardiac defects were not observed with complete prenatal and postnatal diagnostic confirmation during the study period and were therefore not included in the analysis.

Data Collection

Clinical data were collected retrospectively from patient medical records. These data included fetal echocardiography and prenatal ultrasound reports, maternal demographic characteristics, gestational age at diagnosis, the type and classification of the cardiac anomaly, and the presence of associated chromosomal or extracardiac abnormalities. All prenatal imaging was performed by experienced perinatologists using a Samsung V8 high-resolution ultrasound system. When indicated, detailed fetal echocardiographic examinations were performed by pediatric cardiologists.

Perinatal and neonatal data, including birth weight, Apgar scores, gestational age at delivery, mode of delivery, and neonatal outcomes (survival status, need for surgery, prostaglandin E1 infusion, and admission to the neonatal intensive care unit [NICU]), were recorded. Postnatal echocardiographic findings, all evaluated by pediatric cardiologists, were reviewed to assess diagnostic accuracy and confirm the prenatal diagnoses.

Statistical Analysis

Statistical analyses were performed using IBM SPSS Statistics for Mac, version 24.0 (IBM Corp., Armonk, NY, USA). As this was a descriptive study, no comparative or inferential statistical tests were applied. Continuous variables were presented as mean±standard deviation (SD), and categorical variables were expressed as frequencies and percentages.

Results

During the study period, 9,755 fetal echocardiographic examinations were reviewed, of which 92 fetuses with prenatally diagnosed cardiac anomalies were eligible for inclusion. Table 1 summarizes the demographic and clinical characteristics of the study population. The mean maternal age was 29.93±5.37 years, with a mean gravidity of 2.64±1.56 and parity of 1.13±1.05. Assisted reproductive technologies were used in 5 (5.4%) pregnancies, and multiple pregnancies were present in 5 (5.4%) cases.

The mean gestational age at diagnosis of cardiac anomalies was 23.07±4.15 weeks, and the mean gestational age at delivery was 35.56±4.89 weeks. Regarding fetal sex, 49 (53.3%) cases were female and 43 (46.7%) were male.

The mean birth weight was 2868.5±647.6 g. Cesarean delivery was the most common mode of birth, occurring in 61 (73.9%) cases, whereas vaginal delivery occurred in 21 (26.1%) cases. Pregnancy termination was performed in 10 (10.9%) cases due to severe cardiac anomalies and/or associated conditions.

The mean Apgar score at 1 minute was 7.41±0.80, and at 5 minutes, it was 8.60±0.57. A total of 55 newborns (67.0%) required admission to the NICU, with a mean length of stay of 14.30±12.68 days.

Maternal comorbidities were present in 16 (17.3%) patients. The most common condition was gestational diabetes mellitus, observed in 7 cases, followed by type 1 diabetes mellitus in 3 cases. Other conditions included maternal valvular heart disease, epilepsy, hypothyroidism, and preeclampsia.

Extracardiac anomalies were identified in 16 fetuses (17.3%), including pelviectasia, severe hydrocephalus, skeletal dysplasia, alobar holoprosencephaly, dysgenetic corpus callosum, cleft lip and palate, borderline ventriculomegaly, pes equinovarus, gastroschisis, and horseshoe kidney.

Chromosomal or genetic abnormalities were detected in 9 fetuses (9.8%). Five of these fetuses were born and are currently alive, whereas four pregnancies were electively terminated due to severe associated findings.

Septal defects were the most common anomalies, observed in 42 cases (45.7%). Conotruncal defects were diagnosed in 17 cases (18.5%), followed by left-sided obstructive lesions in 14 cases (15.2%) and right-sided obstructive lesions in 11 cases (12.0%). Complex cardiac anomalies were identified in 4 fetuses (4.3%), and isolated anomalies categorized as “other” were observed in 4 cases (4.3%). The distribution of fetal cardiac anomalies by major diagnostic category is presented in Figure 1, while detailed subdiagnostic data are summarized in Table 2.

Table 1. Maternal, fetal, and perinatal clinical characteristics (n=92)

Characteristic	n (%) or Mean±SD
Maternal age (years)	29.93±5.37
Gravidity	2.64±1.56
Parity	1.13±1.05
Abortions	0.52±0.96
IVF pregnancy	5 (5.4)
Multiple pregnancy	5 (5.4)
GA at diagnosis (weeks)	23.07±4.15
GA at delivery (weeks)*	35.56±4.89
Fetal sex (female)	49 (53.3)
Fetal sex (male)	43 (46.7)
Birth weight (g)*	2868.5±647.6
Delivery mode, vaginal*	21 (26.1)
Delivery mode, cesarean section*	61 (73.9)
Pregnancy termination	10 (10.9)
Maternal comorbidities	16 (17.3)
Extracardiac anomalies	16 (17.3)
Genetic/chromosomal anomalies	9 (9.8)
Apgar score at 1 minute*	7.41±0.80
Apgar score at 5 minutes*	8.60±0.57
NICU admission*	55 (67.0)
NICU stay (days)*	14.30±12.68
Postnatal death*	11 (13.4)

*: Calculated among live-born infants (n=82). SD: Standard deviation; IVF: *In vitro* fertilization; GA: Gestational age; NICU: Neonatal intensive care unit.

Of the 92 fetuses, 10 pregnancies were electively terminated due to severe anomalies. Among the remaining 82 live-born infants who underwent postnatal evaluation, prenatal and postnatal cardiac diagnoses were concordant in 72 cases (87.8%), whereas discrepancies were observed in 10 cases (12.2%).

Among the 30 fetuses prenatally diagnosed with ventricular septal defect (VSD), 20 had muscular-type defects (66.7%), including 17 midmuscular and 3 apical VSDs. Two cases (6.7%)

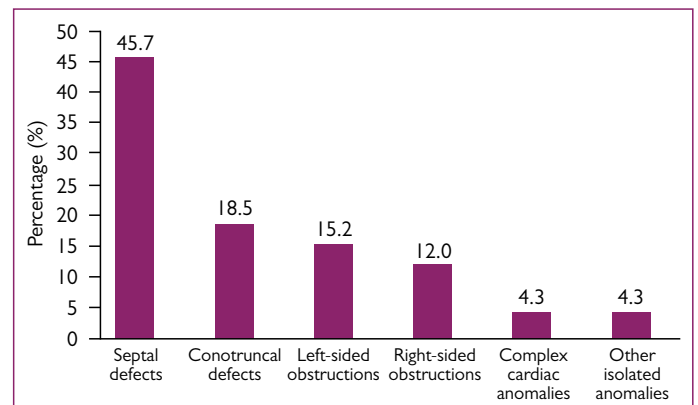


Figure 1. Distribution of fetal cardiac anomalies by major diagnostic category (n=92).

Table 2. Distribution of fetal cardiac anomalies by diagnostic group

Diagnostic group	Diagnosis	n	%
Septal defects	Ventricular septal defect (VSD)	30	32.6
	Complete atrioventricular septal defect (AVSD)	8	8.7
	Partial atrioventricular septal defect (AVSD)	4	4.3
Conotruncal defects	Tetralogy of Fallot	9	9.8
	Transposition of the great arteries (TGA)	4	4.3
	Double outlet right ventricle (DORV)	4	4.3
Left-sided obstructive lesions	Hypoplastic left heart syndrome (HLHS)	8	8.7
	Aortic stenosis	2	2.2
	Coarctation of the aorta	3	3.3
	Tubular aortic hypoplasia	1	1.1
Right-sided obstructive lesions	Hypoplastic right heart syndrome	2	2.2
	Ebstein anomaly	1	1.1
	Pulmonary atresia	3	3.3
	Tricuspid atresia	2	2.2
	Pulmonary stenosis	3	3.3
Complex cases	Hypoplastic right ventricle + tricuspid atresia + TAPVR	1	1.1
	Single ventricle + right isomerism + TAPVR	1	1.1
	Large VSD + tubular aortic hypoplasia	1	1.1
	DORV with transposition + left atrial isomerism + single atrium	1	1.1
Other isolated anomalies	Right aortic arch	4	4.3

TAPVR: Total anomalous pulmonary venous return.

were classified as perimembranous VSDs. Eight prenatally suspected VSDs were not confirmed postnatally.

As shown in Table 3, 11 postnatal deaths were recorded among infants with congenital heart defects. Most cases involved complex structural anomalies and occurred between postnatal day 3 and day 150.

As indicated in Table 4, 10 pregnancies were electively terminated due to severe cardiac anomalies with or without associated extracardiac findings. Cardiac diagnoses included complete atrioventricular septal defect (AVSD) in four cases, hypoplastic left heart syndrome (HLHS) in four cases, and one case each of tetralogy of Fallot and double outlet right ventricle (DORV).

Five fetuses with confirmed chromosomal or genetic abnormalities were carried to term and are currently alive. In one case, tubular aortic hypoplasia was present, and genetic analysis using array comparative genomic hybridization (array-CGH) identified an interstitial deletion at chromosome 6q24–q25. The infant is now one year old and remains under follow-up. Two other cases had partial AVSD associated with Down syndrome, one of whom also had albinism. Another case had a VSD with shortening of the long bones and a postnatal diagnosis of neurofibromatosis, whereas the final case had a VSD accompanied by hydrocephalus and underwent cardiac surgery at three months of age.

Discussion

Congenital cardiac anomalies may be identified during both the prenatal and postnatal periods through systematic ultrasono-

Table 3. Postnatal deaths in fetuses with congenital cardiac anomalies

Postnatal diagnosis	Postnatal day of death
Large VSD + tubular aortic hypoplasia	Day 3
HLHS	Day 4
HLHS	Day 11
Aortic stenosis	Day 14
TGA	Day 16
Complete AVSD	Day 31
HLHS	Day 35
HLHS	Day 37
HRHS	Day 45
DORV with transposition+left atrial isomerism+single atrium	Day 140
Single ventricle+right isomerism+TAPVR	Day 150

HLHS: Hypoplastic left heart syndrome; VSD: Ventricular septal defect; TGA: Transposition of the great arteries; AVSD: Atrioventricular septal defect; DORV: Double outlet right ventricle; HRHS: Hypoplastic right heart syndrome; TAPVR: Total anomalous pulmonary venous return.

graphic evaluation and standardized diagnostic practices. Nevertheless, it is well recognized that a proportion of cardiac defects detected in utero are not confirmed after birth. Previous studies have reported discordance rates between prenatal and postnatal cardiac diagnoses ranging from approximately 10% to 25%, with small septal defects and complex structural anomalies accounting for a substantial proportion of these discrep-

Table 4. Terminated pregnancies due to severe cardiac and/or extracardiac anomalies

Cardiac diagnosis	Associated anomalies	Karyotype	Gestational age (weeks)
Tetralogy of Fallot	Ventriculomegaly, upper limb contracture	Normal karyotype	16
HLHS	Hydrops fetalis	Normal karyotype	24
HLHS	None detected	Normal karyotype	22
HLHS	None detected	Normal karyotype	20
HLHS	None detected	Normal karyotype	21
DORV	Severe hydrocephalus	Normal karyotype	19
Complete AVSD	None detected	Trisomy 21	19
Complete AVSD	Alobar holoprosencephaly	Trisomy 18	21
Complete AVSD	Cleft lip and palate	5p deletion	20
Complete AVSD	Omphalocele and ventriculomegaly	Trisomy 18	19

HLHS: Hypoplastic left heart syndrome; DORV: Double outlet right ventricle; AVSD: Atrioventricular septal defect.

ancies.^[10–13] In a large multicenter cohort study, van Velzen et al.^[10] reported an overall diagnostic accuracy of approximately 85–90% for prenatal echocardiography, while emphasizing that postnatal evaluation remains important for confirming the final diagnosis in certain lesions. Similarly, Bensemlali et al.^[12] demonstrated that differences between prenatal and postnatal findings may occur particularly in complex congenital heart disease and small ventricular septal defects, and that these changes may influence postnatal management strategies.

In the present study, prenatal diagnoses were confirmed postnatally in 87.8% of cases, whereas 12.2% of cases were discordant. This rate is consistent with those reported in earlier series. The majority of discordant findings were attributable to small or muscular ventricular septal defects (VSDs), which accounted for 80% (8 of 10) of the mismatched cases. In addition, one fetus with a prenatal suspicion of coarctation of the aorta showed no postnatal evidence of this lesion, and one case initially diagnosed as tetralogy of Fallot (TOF) was subsequently reclassified as double outlet right ventricle (DORV) after birth.

Several mechanisms may explain the absence of postnatal confirmation of prenatally diagnosed VSDs. These include spontaneous intrauterine closure of small defects, failure to visualize the defect postnatally before closure, or false-positive interpretation during fetal echocardiography.^[14,15] These observations underscore the need for caution when diagnosing small or muscular VSDs in utero and highlight the importance of systematic postnatal cardiologic evaluation in all neonates with suspected prenatal cardiac anomalies.

Fetal cardiac anomalies can be detected from relatively early stages of gestation.^[16] Early identification of major structural abnormalities, particularly those detected toward the end of the first trimester or the beginning of the second trimester, provides a valuable window for advanced genetic testing and informed decision-making regarding pregnancy continuation or termination.^[17] Lugthart et al.^[17] demonstrated that early prenatal detection of severe congenital heart defects is associated with a lower threshold for pregnancy termination, likely due to the increased time available for parental counseling and decision-making. In our study, several pregnancies were electively terminated be-

tween 16 and 24 weeks' gestation because of severe cardiac and/or systemic malformations. These findings emphasize the clinical value of high-resolution imaging systems and experienced multidisciplinary teams in prenatal cardiac diagnosis.

In this cohort, 10.9% of pregnancies were terminated due to severe cardiac defects and/or associated major genetic or extracardiac anomalies. These fetuses frequently exhibited additional systemic abnormalities, including alobar holoprosencephaly, severe hydrocephalus, omphalocele, and limb contractures. Chromosomal abnormalities were identified in four cases, including trisomy 18, trisomy 21, and a 5p deletion. These observations are consistent with previous reports demonstrating that the coexistence of genetic and extracardiac anomalies significantly worsens prognosis and strongly influences pregnancy management decisions.^[18,19]

Accurate perinatal management of prenatally diagnosed congenital heart disease has a direct influence on neonatal outcomes. For critical lesions such as transposition of the great arteries (TGA), hypoplastic left heart syndrome (HLHS), and pulmonary atresia, delivery planning in a tertiary care center with immediate access to neonatal intensive care, pediatric cardiology, and cardiovascular surgery is widely recommended.^[20] This approach allows for the timely initiation of prostaglandin E1 infusion, early respiratory support, and prompt surgical or catheter-based interventions when required.^[6] Although some studies have suggested that prenatal diagnosis does not uniformly reduce postoperative mortality, it appears to improve preoperative hemodynamic stability and early neonatal management.^[21,22]

In the present series, 11 postnatal deaths were recorded, most of which occurred in infants with complex anatomical defects or severe obstructive lesions. Mortality predominantly occurred in the early postnatal period, between day 3 and day 150 of life. These findings indicate that, despite advances in prenatal diagnosis and delivery planning, survival remains limited in a subset of neonates with highly complex cardiac anomalies. Consistent with these observations, previous population-based studies have shown that, although survival among infants with critical congenital heart disease has improved over time, mortality remains substantial, particularly in complex cardiac lesions.^[23]

One of the principal strengths of this study is that all cases were evaluated at a single tertiary referral center using high-resolution ultrasonography and fetal echocardiography systems operated by experienced perinatologists and pediatric cardiologists. The systematic prenatal–postnatal comparison of diagnoses and the detailed assessment of extracardiac and genetic abnormalities enhance the internal consistency and clinical relevance of the findings. Several limitations should also be acknowledged. The retrospective design led to the exclusion of some cases due to incomplete records. In addition, postnatal follow-up is still ongoing for a proportion of patients, precluding a comprehensive evaluation of long-term morbidity and mortality. Finally, the overall number of cases was limited, reflecting the relative rarity of prenatally diagnosed congenital cardiac anomalies. Despite these limitations, the present data provide clinically meaningful insights into diagnostic accuracy and perinatal outcomes in a real-world tertiary care setting.

Conclusion

This study demonstrates that prenatal diagnosis of congenital heart defects plays a central role in guiding perinatal care and postnatal management. The high rate of diagnostic concordance observed in this cohort highlights the clinical value of early and systematic fetal cardiac evaluation, particularly in severe lesions for which delivery planning in specialized centers may favorably influence early neonatal outcomes.

The presence of associated extracardiac and genetic anomalies emerged as a major determinant of prognosis. Accordingly, comprehensive fetal anatomic assessment and formal genetic counseling should be regarded as essential components of care in all pregnancies complicated by suspected fetal heart disease.

Prenatal diagnosis should be viewed not merely as a tool for anomaly detection, but as the foundation for the coordinated management of pregnancy, delivery, and postnatal treatment. Future prospective studies with extended follow-up are needed to better define the long-term clinical implications of this approach and to refine strategies for optimizing outcomes in affected infants.

Disclosures

Ethics Committee Approval: The study was approved by the Koşuyolu High Specialization Training and Research Hospital Scientific Research Ethics Committee (no: 2025/09/1147, date: 03/06/2025).

Informed Consent: Written informed consent was not required due to the retrospective nature of this study.

Conflict of Interest Statement: The authors have no conflicts of interest to declare.

Funding: The authors declared that this study received no financial support.

Use of AI for Writing Assistance: No use of AI-assisted technologies was declared by the authors.

Author Contributions: Concept – F.A.G.; Design – G.E.D.; Supervision – Y.Ç.; Data Collection and/or Processing – A.Y., D.K.; Analysis and/or Interpretation – A.Y., İ.B.; Literature Review – A.Y., B.K.; Writing – A.Y., D.K.; Critical Review – F.A.G.

Peer-review: Externally peer-reviewed.

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