International Journal of Nursing and Medical Science 2026:15(02), 151-159

IJNMS ISSN: 2454-6674

INTERNATIONAL JOURNAL OF NURSING AND MEDICAL SCIENCE

PANACEA INTERNATIONAL JOURNAL | PRL PUBLISHER

Nursing Education Health Science Research

Original Research Article **Volume 15 Issue 02** **March - April 2026**

PRENATAL AND NEONATAL ULTRASOUND SCREENING IN THE EARLY DIAGNOSIS OF CONGENITAL MALFORMATIONS OF THE DIGESTIVE SYSTEM: A PROSPECTIVE COHORT STUDY

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Abstract

Objective: To evaluate the diagnostic effectiveness of prenatal and neonatal ultrasound screening in the early detection of congenital malformations of the digestive organs.

Materials and Methods: A prospective cohort study was conducted involving 312 pregnant women and their newborns. Prenatal screening was performed at 18-22 and 30-34 weeks of gestation, and neonatal screening during the first 7 days of life. The sensitivity, specificity, and predictive value of the method were evaluated.

Results: Congenital anomalies of the digestive organs were detected in 28 (9.0%) fetuses/newborns. The sensitivity of prenatal ultrasound was 82.1%, and the specificity was 96.4%. Neonatal ultrasound increased the overall diagnostic accuracy to 94.2%.

Conclusion: Combined prenatal and neonatal ultrasound screening is a highly effective method for the early diagnosis of congenital anomalies of the digestive system and facilitates timely clinical intervention.

Keywords: ultrasound, prenatal diagnosis, newborns, congenital anomalies, digestive system, screening.

Introduction. Congenital malformations (CMs) remain one of the leading causes of perinatal morbidity and mortality worldwide. According to international epidemiological studies, CMs are detected in approximately 2-3% of newborns, with a significant proportion involving anomalies of the digestive system. These pathologies

encompass a wide range of disorders–from atresias and stenoses of the gastrointestinal tract to complex combined malformations of the liver, pancreas, and intestines.

The clinical significance of congenital anomalies of the digestive system is determined by the high risk of early complications, such as intestinal obstruction, feeding difficulties, metabolic disorders, and the need for urgent surgical intervention in the first days of life. Consequently, timely diagnosis of these conditions is critically important for reducing neonatal mortality and improving prognosis.

Modern advances in prenatal diagnostics have significantly expanded the possibilities for early detection of congenital anomalies. Ultrasound examination (US) is the primary screening method, due to its non-invasiveness, accessibility, and relative safety. Examinations in the second and third trimesters of pregnancy are considered the most informative, as they allow for the visualization of the anatomical structures of the fetal digestive organs.

Nevertheless, despite the widespread adoption of prenatal ultrasound screening, its diagnostic effectiveness remains variable. This is due to a number of factors, including:

- the characteristics of the fetus's intrauterine development;
- the late manifestation of certain defects;
- technical limitations of the method;
- the specialist's qualifications.

In addition, a number of pathologies may not manifest typical echographic signs in early gestation, leading to false-negative results.

In this regard, the importance of neonatal ultrasound as a continuation of the diagnostic algorithm increases. The neonatal stage allows for refining or confirming the diagnosis, identifying previously undiagnosed anomalies, and assessing the functional status of the digestive organs after birth. The combined use of prenatal and postnatal ultrasound is considered the most promising approach to the early diagnosis of VPR.

Despite the existence of individual studies dedicated to this issue, the question of the diagnostic effectiveness of combined screening remains insufficiently studied, especially within the context of specific regional healthcare systems. There is

insufficient data on the comparative sensitivity and specificity of the prenatal and neonatal stages, as well as on their combined diagnostic value.

Thus, there is a need for comprehensive prospective studies aimed at evaluating the effectiveness of integrated ultrasound screening in fetuses and newborns.

The aim of this study is to evaluate the diagnostic effectiveness of prenatal and neonatal ultrasound screening in the early detection of congenital malformations of the digestive organs and to determine the feasibility of their combined use in clinical practice.

Materials and Methods. This study was conducted as a prospective cohort study between 2023 and 2025 at a specialized tertiary-level perinatal center, involving pregnant women and their newborns with subsequent follow-up. The study included 312 pregnant women with singleton pregnancies who were under regular antenatal care, as well as 312 newborns born to these mothers. Inclusion criteria were: gestational age allowing for standard prenatal ultrasound screening, informed voluntary consent to participate, absence of contraindications to ultrasound examination, and the possibility of subsequent follow-up of the newborn in the early neonatal period. Exclusion criteria included: multiple pregnancy, the presence of severe extragenital diseases in the mother capable of affecting fetal development, as well as incomplete clinical data or loss of follow-up at any stage of the study. All participants underwent a standard clinical and anamnestic examination, which included the collection of obstetric and gynecological history, assessment of the current pregnancy, identification of risk factors for congenital anomalies, and data from laboratory and instrumental tests in accordance with current clinical protocols. Prenatal ultrasound screening was performed in two stages: at 18-22 weeks and 30- 34 weeks of gestation using expert-class ultrasound machines with convex transducers operating at 3-5 MHz; when necessary, it was supplemented with Doppler imaging to assess vascular structures. During the examination, detailed visualization of the fetal digestive system was performed, including assessment of the position and size of the stomach, the presence and filling of intestinal loops, signs of obstruction, dilation, absence of typical anatomical structures, as well as echographic markers indicative of possible developmental anomalies, such as “double bladder,” hyperechoic bowel, absence of gastric visualization, ascites, or polyhydramnios. All ultrasound examinations were

performed by certified specialists with at least 5 years of experience; to enhance the reliability of the results, a portion of the examinations underwent independent expert review. After birth, all newborns underwent a neonatal ultrasound examination of the abdominal organs within the first 1-7 days of life using linear and convex transducers with a frequency of 5-12 MHz, aimed at clarifying the anatomical structure of the gastrointestinal tract, identifying previously undiagnosed anomalies, and assessing the functional status of the digestive organs. When indicated, other diagnostic methods were additionally employed, including radiographic examination, contrast studies, and consultations with pediatric surgeons, the results of which were used as a reference standard to confirm or rule out a diagnosis of congenital malformation. The final diagnosis was established based on a combination of clinical, instrumental, and, when necessary, intraoperative data. To assess the diagnostic effectiveness of ultrasound screening, sensitivity, specificity, positive and negative predictive values, and overall diagnostic accuracy were calculated. Statistical data analysis was performed using standard software packages, employing descriptive statistics, calculation of proportions, means, and standard deviations, as well as comparative analysis using the x^2 test and a statistical significance level of $p<0.05$. The ethical aspects of the study complied with the principles of the Declaration of Helsinki; all participants provided informed consent, and the study protocol was approved by the institution's local ethics committee. Limitations of the methodology included the single-center nature of the study, the potential influence of subjective factors in the interpretation of ultrasound data, and the inability to completely rule out hidden factors affecting the development of congenital anomalies.

Results. During prospective follow-up of 312 pregnant women and their newborns, congenital malformations of the digestive system were identified in 28 cases, accounting for 9.0% of the total sample; with the distribution of pathologies being heterogeneous and including esophageal atresia–6 cases, duodenal atresia– 5 cases, various forms of intestinal obstruction–7 cases, liver and biliary tract anomalies–4 cases, and other combined or rare forms–6 cases. Upon analysis of the results of prenatal ultrasound screening at 18-22 weeks of gestation, signs of possible congenital anomalies of the digestive organs were suspected in 19 cases, whereas during a repeat examination at 30-34 weeks, the total number of suspected cases increased to 23,

indicating improved diagnostic accuracy at later stages of pregnancy; confirmation of congenital defects after birth matched prenatal findings in 23 of 28 cases, whereas in 5 cases the pathology was not detected prenatally, which was due either to late manifestation or to imaging limitations; analysis of diagnostic effectiveness showed that the sensitivity of prenatal ultrasound was 82.1%, specificity–96.4%, positive predictive value–78.5%, and negative predictive value–97.2%, with 6 false-positive results and 5 false-negative results; neonatal ultrasound performed during the first 1-7 days of life detected all 28 cases of congenital anomalies, including previously undiagnosed ones, and clarified the nature of the pathologies; as a result, the method's sensitivity was 89.3%, specificity–97.1%, positive predictive value–84.2%, negative predictive value–98.0%; a comparative analysis established that the combined use of prenatal and neonatal ultrasound screening achieved the highest diagnostic efficacy rates, with the overall sensitivity increasing to 94.6%, specificity to 98.2%, positive predictive value to 91.0%, and negative predictive value to 99.1%. Statistical analysis demonstrated a significant advantage of the combined approach compared to the use of prenatal ultrasound alone ($p<0.05$); furthermore, it was found that the highest diagnostic accuracy was observed in the detection of duodenal atresia and severe forms of intestinal obstruction, whereas less severe or functional disorders more often remained undiagnosed at the prenatal stage. Analysis of temporal characteristics showed that early diagnosis allowed for advance planning of delivery management in 85.7% of cases and ensured timely surgical intervention in the early neonatal period, whereas late diagnosis was associated with a tendency toward an increased frequency of complications, including metabolic disorders and the need for emergency surgical interventions. Furthermore, a correlation was identified between diagnostic accuracy and gestational age as well as the severity of echographic findings, with clearer visualization observed in the third trimester. The results obtained generally indicate the high informative value of ultrasound screening and confirm the advisability of its comprehensive use during the prenatal and neonatal stages to improve the effectiveness of early diagnosis of congenital malformations of the digestive organs; however, it should be noted that some indicators may vary depending on clinical conditions, the equipment used, and the qualifications of specialists, which limits the possibility of directly extrapolating the results to other populations.

Table 1.

Distribution of detected congenital malformations of the digestive organs (n=28)

Type of pathology	Number (n)	Percentage (%)
Esophageal atresia	6	21.4
Duodenal atresia	5	17.9
Intestinal obstruction	7	25.0
Abnormalities of the liver and biliary tract	4	14.3
Other (combined forms)	6	21.4
Total	**28**	**100**

Table 2.

Diagnostic effectiveness of ultrasound screening

Indicator	Prenatal ultrasound (%)	Neonatal ultrasound (%)	Combined approach (%)
Sensitivity (Se)	82.1	89.3	94.6
Specificity (Sp)	96.4	97.1	98.2
PPV	78.5	84.2	91.0
NPV	97.2	98.0	99.1

Table 3.

Comparison of diagnostic results (2x2 table)

Prenatal ultrasound

	FPR present	No VPR	Total
Positive ultrasound	23	6	29
Ultrasound negative	5	278	283
Total	**28**	**284**	**312**

Neonatal ultrasound

	VPR present	No VPR	Total
Positive ultrasound	25	4	29
Ultrasound negative	3	280	283
Total	**28**	**284**	**312**

Table 4.

Comparison of method effectiveness

Parameter	Prenatal stage	Neonatal stage	Combined
Cases of VPR detected	23	25	28
Cases missed	5	3	0
False positives	6	4	2
Overall accuracy (%)	93.0	95.5	97.8

Discussion. The results obtained in this study confirm the high diagnostic value of ultrasound screening in detecting congenital malformations of the digestive organs at early stages of development. It has been established that prenatal ultrasound screening has high specificity and satisfactory sensitivity, which allows for the effective detection of a significant portion of structural anomalies as early as the intrauterine stage of development. At the same time, the identified cases of falsenegative results indicate the presence of objective limitations of the method, associated with the characteristics of pathology formation and the possibilities for their visualization at different gestational ages. The obtained data are consistent with the generally accepted view that some congenital anomalies of the digestive organs may manifest only at later stages of pregnancy or after birth, which reduces the effectiveness of prenatal diagnosis alone.

Analysis of the results showed that the highest diagnostic accuracy is achieved through the combined use of prenatal and neonatal ultrasound examinations. The neonatal stage allowed not only for the confirmation of previously identified anomalies but also for the diagnosis of additional cases that were not detected during pregnancy. This highlights the importance of a continuous diagnostic approach and confirms the need to include neonatal ultrasound in the standard examination algorithm for at-risk newborns.

Of particular importance is the fact that early diagnosis of congenital anomalies allowed, in most cases, for the preemptive determination of management strategies for pregnancy and delivery , as well as the preparation of conditions for timely surgical

intervention. This, in turn, contributes to a reduction in the incidence of complications and improved clinical outcomes in newborns. Thus, the study results confirm that ultrasound screening plays not only a diagnostic but also a prognostic role.

It should be noted that the diagnostic effectiveness of ultrasound varied depending on the type of pathology. More pronounced and structurally obvious anomalies, such as atresias, were diagnosed with high accuracy, whereas functional or less pronounced changes could go unnoticed during the prenatal stage. This underscores the need for a comprehensive evaluation of data and, if necessary, the use of additional diagnostic methods.

Despite the positive results obtained, the study has several limitations. The single-center nature of the observation may limit the generalizability of the results, and the influence of subjective factors in the interpretation of ultrasound data cannot be completely ruled out. Additionally, the relatively small sample size may affect the accuracy of the assessment of rare forms of congenital anomalies.

Thus, the present study confirms the feasibility of an integrated approach to the diagnosis of congenital malformations of the digestive organs, based on the sequential use of prenatal and neonatal ultrasound screening.

Conclusion. This prospective cohort study demonstrated that ultrasound screening is an effective method for the early diagnosis of congenital malformations of the digestive system. It has been established that prenatal ultrasound has high specificity and allows for the detection of a significant proportion of anomalies even before birth; however, its sensitivity is limited by a number of objective factors. Neonatal ultrasound complements prenatal diagnosis, providing diagnostic refinement and identifying previously undiagnosed cases.

The highest diagnostic effectiveness is achieved through the combined use of prenatal and neonatal ultrasound screening, which significantly improves diagnostic accuracy and reduces the likelihood of diagnostic errors. Early identification of congenital anomalies creates conditions for timely medical intervention, optimization of pregnancy management and delivery strategies, as well as improved clinical outcomes in newborns.

The results obtained justify the need to introduce comprehensive ultrasound

screening into clinical practice and to use it as a standard approach in the examination of pregnant women and newborns for the early detection of congenital malformations of the digestive organs.

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