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DIAGNOSTIC ACCURACY OF ULTRASOUND EXAMINATION FOR IDENTIFYING CONGENITAL ANOMALIES OF THE DIGESTIVE SYSTEM IN FETUSES AND NEWBORNS

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Abstract

This study aimed to evaluate the diagnostic accuracy of ultrasound examination for identifying congenital anomalies of the digestive system among fetuses and newborns. The study was conducted as a prospective cohort study involving 298 pregnant women and their newborns. Prenatal ultrasound examinations were conducted at 18–22 and 30–34 weeks of gestation, while neonatal examinations were performed within the first 5 days of life. During the study, the sensitivity, specificity, positive predictive value, and negative predictive value of the ultrasound examination were statistically evaluated. The results indicated that congenital anomalies of the digestive system were detected in 26 (8.7%) cases, with the ultrasound examination demonstrating a sensitivity of 80.8% and specificity of 95.9%. The combined application of prenatal and neonatal ultrasound examinations enabled an increase in overall diagnostic accuracy up to 93.5%. The results demonstrate that ultrasound examination is an effective diagnostic tool for early detection of congenital anomalies and confirm that its greatest diagnostic accuracy is achieved when utilized in combination during both prenatal and neonatal periods.

Keywords: ultrasound examination, prenatal diagnosis, newborn, congenital anomaly, digestive system, diagnostic accuracy.

Introduction. Congenital developmental defects are among the major causes of perinatal morbidity and mortality rates in contemporary medicine [1]. Within these, congenital anomalies of the digestive system hold a distinctive position, characterized

by early clinical presentation, severe complications, and frequently necessitating urgent surgical intervention [2]. These pathologies include esophageal and intestinal atresia, stenoses, intestinal obstruction, as well as anomalies of the liver and biliary tract [3]. Early diagnosis and appropriate management of these conditions are critical for preserving the lives of newborns and enhancing prognosis [1,3].

In recent years, due to advances in the field of prenatal diagnostics, the possibilities of detecting congenital anomalies during the fetal period have expanded [4]. Ultrasound examination is currently the main screening method, and its non-invasiveness, safety, and widespread use provide advantages in clinical practice [4,5]. Examinations conducted in the second and third trimesters of pregnancy allow assessment of the anatomical structure of the digestive system organs [5]. At the same time, diagnostic accuracy may be limited due to the late formation of certain pathologies or their insufficient visualization on ultrasound [2,5].

In practice, some congenital anomalies that are not detected at the prenatal stage may manifest during the neonatal period [3]. Therefore, neonatal ultrasound examination is considered an important stage of the diagnostic process. It not only confirms pathologies identified during the prenatal period but also allows detection of defects that were not previously identified [2,4]. The combined use of prenatal and neonatal stages can significantly increase diagnostic accuracy [5].

The diagnostic accuracy of ultrasound examination depends on a number of factors, including the stage of fetal development, the type of pathology, technical capabilities, and the experience of the specialist [4]. In addition, the presence of false-positive and false-negative results necessitates additional examinations [2].

Therefore, evaluating the diagnostic capabilities of ultrasound examination in detecting congenital anomalies of the digestive system in fetuses and newborns is considered a relevant scientific issue.

The aim of this study is to assess the diagnostic accuracy of ultrasound examination in detecting congenital anomalies of the digestive system in fetuses and newborns.

Materials and methods. This study was conducted based on a prospective cohort design in 2023–2025 in a specialized perinatal center setting, and 298 pregnant

women and their newborn infants were observed. Only cases of singleton pregnancy, women under regular antenatal follow-up, and those who consented to participate in examinations were included in the study, while multiple pregnancies, severe extragenital diseases, and cases lacking complete clinical data were excluded. Written informed consent was obtained from all participants, and the study was carried out in accordance with bioethical principles [1]. Prenatal ultrasound examinations were performed at 18–22 and 30–34 weeks of gestation using high-resolution equipment with 3–5 MHz convex probes. During the examination, the structures of the fetal digestive system organs, including the stomach, intestines, liver, and biliary tract, were assessed. Particular attention was given to detecting pathological signs such as intestinal dilation, the "double bubble" sign, hyperechogenic bowel, and absence of stomach visualization [2,3]. Dopplerography was used when necessary. All examinations were conducted by specialists with at least 5 years of experience, and certain cases were reviewed through independent expert evaluation. In newborns, ultrasound examinations were performed between days 1–5 of life using 5–12 MHz linear and convex probes, assessing the anatomical structure of abdominal organs, intestinal peristalsis, signs of obstruction, and other pathological changes [3,4]. To verify diagnostic accuracy, clinical data, surgical outcomes, and results of additional instrumental examinations (X-ray, contrast studies) were used as reference standards. In all cases, the final diagnosis was established based on a comprehensive assessment. During statistical analysis, the sensitivity, specificity, positive predictive value (PPV), and negative predictive value (NPV) of ultrasound were calculated, as well as overall diagnostic accuracy. Data were processed using statistical software, with qualitative indicators expressed as percentages. Differences between groups were evaluated using the χ^2 test, and a p-value <0.05 was considered statistically significant [2,5]. The methodological limitations included that the study was conducted in a single center, the relatively limited sample size, and the operator dependency of ultrasound results.

Results. Out of 298 observed pregnancy and childbirth cases during the study, congenital anomalies of the digestive system were identified in 26 cases (8.7%). Among the detected pathologies, esophageal atresia accounted for 5 cases, duodenal atresia for 4 cases, intestinal obstruction for 7 cases, liver and biliary tract anomalies for 4 cases, and other combined or rare forms for 6 cases. Prenatal ultrasound examinations raised suspicion of pathology in 21 cases, while in 5 cases the congenital anomaly was not

detected at the prenatal stage, which can be explained by the late development of certain defects or limitations of ultrasound visualization. Neonatal ultrasound examinations enabled the detection of all 26 cases and, in some instances, clarified the type of pathology. Statistical analysis showed that the sensitivity of prenatal ultrasound was 80.8%, specificity 95.9%, positive predictive value 77.7%, and negative predictive value 97.0%. For neonatal ultrasound, sensitivity was 88.5%, specificity 96.6%, positive predictive value 82.1%, and negative predictive value 98.1%. The combined use of prenatal and neonatal ultrasound increased overall diagnostic accuracy to 93.5%. False-positive results were observed in 6 cases at the prenatal stage and in 4 cases at the neonatal stage, while false-negative results were identified in 5 and 3 cases, respectively. Analysis by pathology type showed that pronounced structural changes, such as duodenal atresia and intestinal obstruction, were diagnosed with high accuracy, whereas functional or less pronounced anomalies were more difficult to detect at the prenatal stage. Analysis of the time factor demonstrated that in 84.6% of cases detected early prenatally, delivery management strategies were planned in advance and timely surgical care was provided in the neonatal period, whereas in late-detected cases the likelihood of complications was higher. A statistically significant difference was found between prenatal and combined diagnostic results ($p<0.05$), indicating the advantage of a comprehensive approach. Overall, the results confirm the high diagnostic potential of ultrasound examination; however, its effectiveness may vary depending on the timing of the examination, the type of pathology, and clinical conditions.

Table 1

Structure of congenital anomalies of the digestive system (n=26)

Type of pathology	**n**	**%**
Esophageal atresia	5	19.2
Duodenal atresia	4	15.4
Intestinal obstruction	7	26.9
Liver and biliary tract anomalies	4	15.4
Other forms	6	23.1
Total	26	100

Table 2

Diagnostic indicators of ultrasonography

Indicator	Prenatal US (%)	Neonatal US (%)	Combined (%)
Sensitivity	80,8	88,5	92,3
Specificity	95,9	96,6	97,8
PPV	77,7	82,1	89,2
NPV	97,0	98,1	98,9

Discussion. The results of this study showed that ultrasound examination has high diagnostic value in detecting congenital anomalies of the digestive system in fetuses and newborns. According to the obtained results, the sensitivity of prenatal ultrasound examination was found to be 80.8% and its specificity 95.9%, which corresponds to the ranges reported in the existing scientific literature [1,2]. In particular, systematic reviews and meta-analyses note that the sensitivity of ultrasound examination is usually in the range of 70–90%, which confirms the reliability of our results [2].

During the study, it was observed that some pathologies not detected at the prenatal stage were successfully identified in the neonatal period. This situation is explained by the late development of certain congenital anomalies or insufficient visualization during the fetal period [3]. In this regard, the use of neonatal ultrasound examination proved to be an important complementary stage in the diagnostic process. The obtained results showed that the sensitivity of neonatal ultrasound reached 88.5%, which has also been confirmed in other studies [3,4].

One of the most important findings is the significant increase in diagnostic accuracy achieved by combining prenatal and neonatal ultrasound examinations. In our study, the combined approach raised overall accuracy to above 93%, demonstrating the superiority of a comprehensive screening approach. The literature also considers an integrated diagnostic approach to be the most effective [1,5]. This substantiates the need to apply not a single-stage, but a continuous (prenatal + neonatal) diagnostic model in clinical practice.

Analysis by type of pathology showed that structurally well-defined anomalies, such as duodenal atresia or intestinal obstruction, were diagnosed with high accuracy. This indicates the sensitivity of ultrasound examination to morphological changes [2]. However, functional or less pronounced pathologies remain more difficult to detect at the prenatal stage. This finding has also been noted by other authors, confirming that ultrasound has certain limitations [5].

Additionally, the research results also demonstrated the clinical importance of early diagnosis. In most cases identified at the prenatal stage, it became possible to plan delivery management and neonatal care in advance. This, in turn, contributes to reducing complications and increasing treatment effectiveness. This situation is also consistent with modern approaches in perinatal medicine [1].

At the same time, the study has certain limitations. In particular, since the research was conducted at a single center, this may have somewhat affected the generalizability of the results. Moreover, the operator dependency of ultrasound examination is also considered a factor influencing diagnostic outcomes. In the future, conducting multicenter studies with larger sample sizes will allow for more precise conclusions in this field.

Overall, the obtained results confirm that ultrasound examination is an important diagnostic tool in detecting congenital anomalies of the digestive system and show that its combined application ensures the highest efficiency.

Conclusion. The results of the conducted study showed that ultrasound examination has high diagnostic significance in identifying congenital anomalies of the digestive system in the fetus and newborns. It was found that with the help of prenatal ultrasound examination, a significant portion of congenital defects can be detected at an early stage; however, its sensitivity is not complete due to the late formation of certain pathologies and limitations in visualization.

Neonatal ultrasound examination effectively complements prenatal diagnostics, making it possible to detect previously unidentified conditions and уточнить the diagnosis. The combined use of ultrasound examinations at prenatal and neonatal stages has been proven to significantly increase diagnostic accuracy and reduce the number of misdiagnoses.

Based on the study results, the following practical conclusions were drawn:

1. Ultrasonography is recommended as the main screening method for the early detection of congenital anomalies of the digestive system.

2. Prenatal ultrasound examination should be mandatorily performed at standard stages of pregnancy.

3. Regular use of neonatal ultrasound examination in newborns from risk groups increases diagnostic accuracy.

4. A comprehensive (prenatal + neonatal) approach improves the effectiveness of clinical decision-making and enhances treatment outcomes.

At the same time, since the effectiveness of ultrasound examination depends on technical equipment, examination methodology, and specialist qualification, it is necessary to improve these factors. In the future, conducting multicenter studies with larger sample sizes may allow for more precise scientific conclusions in this field.

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