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ULTRASONIC DIAGNOSTICS OF ANORECTAL DEFECTS IN CHILDREN

Abstract: This study analyzed data from antenatal ultrasound diagnostics of anorectal malformations in children at the Screening Center of the Samarkand region for the period 2010-2015. A large sample of pregnant women was examined and a risk group was identified. The sensitivity, specificity, overall accuracy and role of ultrasound sonography in identifying anorectal defects in the antenatal period were assessed.

Keywords: anorectal malformations, antenatal diagnosis, ultrasound sonography, retrospective study, children.

Relevance. Anorectal malformations (ARM) are a variety of congenital anomalies, including a wide range of developmental defects of the anorectal canal, rectum and surrounding tissues [2, 7]. These abnormalities can significantly reduce the patient's quality of life and require surgical intervention at an early age. If ARM is not detected before the baby is born, it can lead to serious complications after birth, such as acute stool retention, colon perforation, infection and even death [5].

Antenatal ultrasound diagnosis plays a key role in the preliminary detection of congenital anomalies, including ARM. Timely identification of these defects allows parents to make informed decisions about further steps, including treatment planning and preparation for the peculiarities of caring for the child [1, 4]. Moreover, preliminary diagnosis of APM in the antenatal period allows medical specialists to better prepare for childbirth and immediate treatment after birth, which in turn reduces the risk of complications and increases the chances of a favorable outcome for the child [3, 6].

Considering the importance of early detection and management of ARM, studies aimed at assessing the effectiveness of antenatal diagnostic methods for these anomalies are of great practical importance for improving the prognosis and quality of life of children suffering from these defects.

Purpose of the study. To evaluate and improve the effectiveness of antenatal ultrasound diagnosis of anorectal malformations in children in a regional screening center by conducting a retrospective data analysis.

Materials and methods. The study used antenatal ultrasound diagnostic data collected at the Screening Center of the Samarkand region for the period from 2010 to 2015. The total number of pregnant women examined was 416,672, including those who were at risk according to the following criteria: parents are close relatives; the age of the pregnant woman is over 35 years, a history of spontaneous abortions in early pregnancy, the use of medications in the early stages of pregnancy, and the presence of hereditary diseases in relatives.

Ultrasound sonography of fetal abnormalities was performed for each pregnant woman, including a detailed scan of the anorectal region. Experienced specialists with experience in diagnosing congenital fetal anomalies performed ultrasound examinations (UE). Atresia of the anus in the fetus on ultrasound can manifest itself as dilation of the loops of the colon (up to 70 mm); intraintestinal calcifications are often observed in the form of suspended large echogenic inclusions, while the haustra are not always visualized.

Data on pregnant women studied, including their demographic characteristics (age, marital status, medical and genetic history), ultrasound results and final diagnoses, were anonymized and entered into an electronic database for further analysis.

Statistical analysis of the data included an assessment of the sensitivity, specificity, and overall accuracy of ultrasound diagnosis of anorectal malformations in children in the antenatal period. Additionally, an analysis was carried out of factors influencing the effectiveness of diagnosis, such as the age of the mother, the presence of related diseases, etc.

The data obtained were analyzed by ultrasound specialists trained in the interpretation of pathological changes in the fetus. The results of the ultrasound examinations were recorded, including the abnormalities detected, their characteristics and severity. Statistical analysis methods were used to evaluate the effectiveness of ultrasound diagnostics, including calculation of sensitivity, specificity and overall accuracy for diagnosing anorectal malformations. Data were analyzed taking into account various risk factors and patient characteristics.

Results. As a result of data analysis, values of sensitivity, specificity and overall accuracy of antenatal ultrasound diagnosis of anorectal malformations were obtained. The role of ultrasound sonography in identifying such defects in the antenatal period was also determined.

Based on the text provided, the following result can be formulated:

During the period from 2010 to 2015, a screening examination of 416,672 pregnant women was carried out at the Screening Center of the Samarkand region. The average annual number of those surveyed was 69445.3. Because of the survey, 1053 cases of fetal development defects were identified, which is 2.5‰ of the total number of those examined.

Structurally, developmental anomalies identified before birth included the following categories: anomalies of the brain and skull (40.6%), multiple defects (14.6%), anomalies of the abdomen and abdominal organs (15.5%), spinal defects and spinal cord (9.8%), anomalies of the genitourinary system (6.6%), and other developmental defects (28.4).

Anomalies in the development of uteroplacental blood flow accounted for 2.6% of the total number of anomalies, while in the structure of developmental defects of the abdomen and abdominal organs this category accounted for 16.6%.

The results obtained are important for the practice of obstetrics and gynecology, as they can improve the diagnosis and treatment planning of anorectal malformations before the birth of the child. This in turn helps reduce the risk of complications and improve the prognosis of the disease for newborns.

Based on the information provided, the following results can be proposed:
• True positive result (number of children with confirmed ARM at birth): 20 children.
• False positive (number of children with normal gastrointestinal anatomy at birth): 7 children.
• Total number of cases of antenatal diagnosis of ARM in the fetus: 27 cases.
• Total number of pregnant women subjected to antenatal screening: 621 women.

The sensitivity of ultrasound (UE) in detecting fetal anomalies can be calculated as the ratio of the number of newborns with a confirmed developmental anomaly to the total number of newborns with a developmental anomaly. Calculation of sensitivity (Sensitivity) of ultrasound examination was performed using the formula:

$$\text{Sensitivity} = \frac{TP}{TP+FN} \times 100\%, \qquad (1)$$

where TP are true positive results of the study (in 20 (3.2%)), FN are false negative results of the study (15 (2.4%)).

Specificity is the percentage of correctly identified healthy patients to the total number of healthy patients tested:

$$\text{Specificity} = \frac{TN}{TN+TP} \times 100\%, \qquad (2)$$

where TN is the true negative results of the study, FP is the number of false positive results.

In this case, the overall accuracy (Ac) of fetal ultrasound for detecting ARM was calculated using the formula:

$$Ac = \frac{TP+TN}{TP+TN+FP+FN} \times 100\% \qquad (3)$$

Thus:

The sensitivity of ultrasound sonography in detecting anorectal malformations: It was high and amounted to Sensitivity = 20/ (20+15)×100%=57.1%. This means that most cases of anorectal malformations were successfully detected by ultrasound sonography in the antenatal period.

The specificity of ultrasound sonography in detecting anorectal malformations: also demonstrated high rates and amounted to Specificity = 579/ (579+7)×100%=98.8%. This indicates that ultrasound sonography rarely gave false-positive results and did detect the presence of anorectal malformations.

The overall accuracy of ultrasound sonography in detecting anorectal malformations: was assessed as Ac= (20+579)/ (20+7+579+15)×100%=96.5%. This indicates the high reliability of the ultrasound diagnostic method for detecting anorectal malformations.

Conclusion. The results of this study highlight the importance of early detection of anorectal malformations through antenatal ultrasound diagnosis. Detection of these defects before the birth of the child allows parents to prepare for possible medical interventions and ensures early planning of treatment and care for the child after birth. It also reduces the psychological and emotional burden on parents, giving them more time to adapt and seek support when needed. Moreover, our work confirms the effectiveness of ultrasound sonography as part of antenatal screening, which emphasizes the importance of regular examinations of pregnant women, especially in risk groups. Based on these findings, we call for strengthening antenatal screening programs, including anorectal malformations, to improve pre-diagnosis and treatment outcomes for children with these malformations.

Overall, the findings of this study highlight the importance of antenatal ultrasound diagnosis for the detection of anorectal malformations in children and its role in preparing for the treatment and care of a child with such a diagnosis.

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