НЕОНАТОЛОГІЯ, ХІРУРГІЯ ТА ПЕРИНАТАЛЬНА МЕДИЦИНА
NEONATOLOGY, SURGERY AND PERINATAL MEDICINE
Т. XIV, № 4(54), 2024
VOL. XIV, № 4(54), 2024
ISSN 2226-1230 (PRINT) ISSN 2413-4260 (ONLINE)
KEY TITLE: NEONATOLOGÌÂ, HÌRURGÌÂ TA PERINATAL'NA MEDICINA (ONLINE)
ABBREVIATED KEY TITLE: NEONATOL. HÌR. PERINAT. MED. (ONLINE)

UDC: 617.7-007.681-053: 616-073.582: 616-08-039.11
DOI: 10.24061/2413-4260. XIV.4.54.2024.29

ULTRASOUND AS A SCREENING TOOL FOR FETAL GLAUCOMA: A CLINICAL APPROACH

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Summary

The World Health Organization's VISION 2020 initiative has recognised paediatric glaucoma as the third leading cause of significant visual impairment and blindness in children in low- and middle-income countries, accounting for 5 % of global childhood blindness. Childhood glaucoma is a critical condition in paediatric ophthalmology, causing damage to the optic nerve and ganglion cells.

***The objective of the study** was to assess the feasibility of ultrasonography screening in pregnant women for the early identification of probable congenital glaucoma in the fetus.*

***Material and methods of the study.** To achieve this, we conducted a comprehensive ultrasound examination of 500 pregnant women at the Republican Centre for Mother and Child Screening in 2022-2024. The article was prepared on the basis of the results of the planned dissertation research (Registration code B2023.3.DSc/Tib902; due dates: 2022-2025).*

***Results:** Ultrasound scans of pregnant women provide important information for ophthalmologists. Ultrasound screening of pregnant women, monitoring the orbits and eyeballs of the fetuses, was performed from 20 to 40 weeks of gestation. Of the total, data were analysed for 2 (0.4 %) cases of fetal eye pathology in which echographic indicators of glaucoma, known as buphthalmos, were identified. After birth, the suspected diagnosis of congenital glaucoma was confirmed in the children, requiring emergency hospitalisation and surgical intervention. This is particularly important in children with congenital glaucoma to prevent blindness and visual impairment in the future.*

***Conclusions:** This imaging could serve as a cost-effective and safe screening technique that provides important information to ophthalmologists. Comprehensive ultrasound assessment of embryonic ocular structures may provide significant information and identify congenital glaucoma in infants. Accurate diagnosis and verification of congenital glaucoma typically requires more specialised ophthalmological assessment postnatally.*

***Key words:** Congenital glaucoma; Fetal Ultrasound; Screening.*

Relevance

The World Health Organization's VISION 2020 initiative has recognised paediatric glaucoma as the third leading cause of significant visual impairment and blindness in children in low- and middle-income countries, accounting for 5 % of global childhood blindness [1, 2]. Childhood glaucoma is a critical condition in paediatric ophthalmology, causing damage to the optic nerve and ganglion cells [3, 4]. The incidence of paediatric glaucoma varies in different populations. For example, the estimated annual incidence rate of paediatric glaucoma in patients aged <20 years at presentation is 0.92 per 100,000 population in Hong Kong [5]. In a series in the United States, the reported incidence was 2.29 per 100,000, or 1 per 43,575 population younger than 20 years [6], and a higher incidence was found in Slovakian gypsies (1/1250) and in Saudi Arabia (1/2500) [7]. Glaucoma is a disease characterised by elevated intraocular pressure leading to permanent vision loss [3, 8, 9]. The prognosis of glaucoma is largely dependent on early detection, effective surgical intervention and management of elevated intraocular pressure (IOP) [2, 10, 12, 12, 13]. Delayed diagnosis of paediatric glaucoma can lead to blindness and cause emotional and psychological distress to the patient's caregivers. A high index of suspicion by paediatricians or general practitioners and prompt referral to an ophthalmologist for a complete eye examination are essential for the timely diagnosis of glaucoma in children [14, 15, 16]. More effective screening strategies may be beneficial for early diagnosis and treatment.

Fetal ocular ultrasound is not yet a significant part of prenatal diagnosis. In general, national and international guidelines only cover the presence of orbits and lenses. However, in recent years, with the development of high-resolution ultrasound equipment and advances in prenatal medicine and genetics, careful examination of the fetal eye has made it possible to diagnose many ocular anomalies before birth [17, 18, 19].

Although screening ultrasound can help to identify potential symptoms of congenital glaucoma, it is not usually the main diagnostic test for this condition.

Objective of the study: The purpose of this study is to investigate whether or not ultrasonography screening of pregnant women could be used to detect possible cases of congenital glaucoma in the developing fetus at an earlier stage.

Material and methods of the study

We conducted a comprehensive examination of 500 pregnant women at the Republican Center for Screening of Mother and Child for the period 2022-2024 on the Samsung WS80A ultrasound diagnostic device with wide-band C5-2 sensors with a frequency range of 2.0-5.0 MHz and 3D8-4 with a frequency range of 4.0-8.0 MHz.

The following criteria were used to determine which pregnant women were eligible for the study:

– hereditary predisposition (glaucoma in a parent or relative)

– a history of giving birth to children with glaucoma

– women who have had infectious diseases during pregnancy (rubella, measles, influenza and other viral infections)

– heavy pregnancy with various pathologies (polyhydramnios)

– malnutrition before and during pregnancy

– severe toxicosis during pregnancy

The examinations were carried out with the help of an expert in ultrasound diagnostics specialising in the highest category. In both the axial and coronal planes, the eyes of the foetuses were examined for their characteristics. In the axial plane, scans were taken from the top of the skull down the face of the foetus. In the coronal plane, the focus of the scan was moved from the tip of the nose to the back of the eye. Both scans included images of the eyelids, cornea, anterior chamber, lens, sclera, iris, hyaloid artery, retina and optic nerve. The dimensions of the eyeball and the positions of the orbits, hyaloid artery and lens were carefully considered. Transvaginal sonography was used in cases where the fetus was in the occipital presentation. This allowed more accurate imaging of the many structures that make up the eye.

The article was prepared on the basis of the results of the planned dissertation research (Registration code B2023.3.DSc/Tib902; due dates: 2022-2025).

Results of the study

The visual prognosis of these patients depends on the timing of presentation and treatment. Delays in presentation, diagnosis and treatment can lead to devastating visual outcomes. Delayed presentation is mainly due to lack of awareness of the disease among caregivers and clinicians. A large tertiary centre study in South India showed that almost half of the childhood glaucoma cases had delayed presentation to the tertiary centre by more than three months from the time the caregivers recognized symptoms, although most parents or caregivers recognized symptoms within the first week after birth [7, 20, 21, 22].

To assess the basic anatomy of the fetus, ultrasound examinations of the fetal orbit and eyes were performed as early as 11-14 weeks of gestation. After 20 weeks of gestation, further detailed anatomical screening was typically performed. The fetal orbit was observed to be an anechoic (echo-transparent) region located symmetrically on either side of the fetal nose (see Figure 1). Between 30 and 36 weeks of gestation, the sagittal diameter of the normal globe is 14.48±0.50 mm, while at 36 weeks it is 16.08±0.13 mm. The fetal lens was found to be a hyperechoic ring with a spherical shape and an anechoic core.

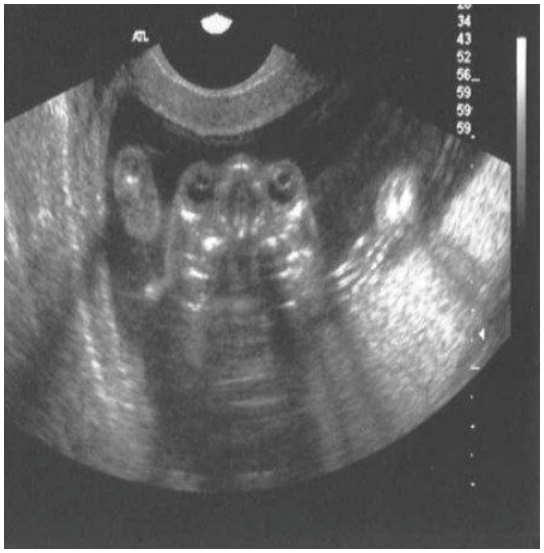

Fig. 1. Coronal ultrasound image showing normal orbital area

Ultrasound was used to evaluate the size of the eye by measuring the anterior-posterior axis (APA) of the eyeball, along with an assessment of the anatomical condition of the eyeballs. Changes in these parameters may indicate a possible diagnosis of glaucoma.

Of the total sample, data were analyzed from 2 (0.4 %) pregnant women with fetal ocular pathology characterized by sonographic indicators of glaucoma, specifically buphthalmos, which is defined by an increase in the size of the eye. The ultrasound screening of pregnant women, along with monitoring of fetal ocular status, was performed between 20 and 40 weeks of gestation.

A 20-year-old primigravida undergoing systemic steroid therapy for chronic nephritis underwent a screening ultrasound at 30 weeks' gestation, which revealed bilateral buphthalmos (OU). This condition had previously been noted at 18 weeks' gestation with evidence of an enlarged anteroposterior diameter of the globe. At 35 weeks' gestation, OU axial length measurements ranged from 23 to 25 mm (Fig. 2), with an approximate measurement of 24 mm derived from B-scan images.

A volume discrepancy between the eye and the orbit was observed; however, no other abnormal changes were noted on the images.

A male infant was delivered vaginally at 38 weeks gestation. The patient weighed 2730 g and measured 48.8 cm in length at birth. The recorded Apgar score was 9 points. The infant was referred to the ophthalmology department of the Tashkent Pediatric Medical Institute (TashPMI) with suspected congenital glaucoma in both eyes. The mother of the newborn visited the doctor on the twelfth day after delivery. The child was diagnosed with OU – primary congenital glaucoma, advanced stage, uncompensated. Keratopathy refers to any disease or disorder affecting the cornea of the eye. An examination under anesthesia and surgical treatment, specifically antiglaucoma surgery, was recommended.

ISSN 2226-1230 (PRINT) ISSN 2413-4260 (ONLINE)

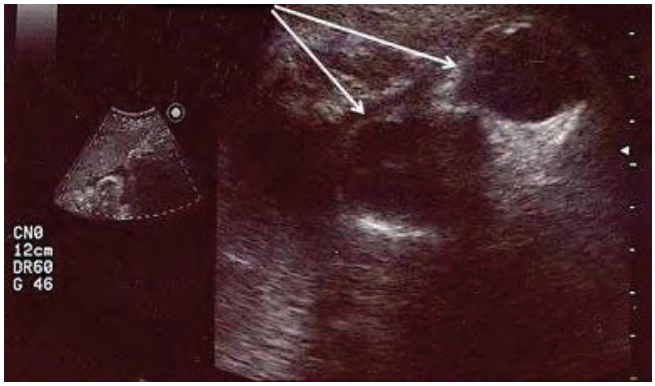

Fig. 2. Echotomographic images of the fetus at 30 weeks of gestation, an increase in the size of the eyeball is noted (indicated by arrows)

Status localis. The eyeballs have a spherical morphology and are enlarged in size. The cornea has an increased diameter, is edematous and shows Haab's striae. The limbus has an increased diameter. The sclera has a white appearance with a bluish tinge. The anterior chamber is deep and the fluid is clear. The pupil is central, round and black. Response to light is minimal. The iris has a smooth pattern and relief. The lens is transparent. The pink reflex of the fundus is not visible due to corneal edema. OU = (digital tension) +1.0.

On day 17, the child was examined under anesthesia and underwent anti-glaucoma surgery.

Tonometry and tonography data according to Friedenwald.

P t= 30 mm Hg / 29 mm Hg

R o = 24.38/25.72

C=0.04/0.09

F = 0.65/1.49

BK (Baker's coefficient)=541.97/272.27

APA (antero-posterior axis) OD=24.2 mm OS=23.5 mm.

Keratometry: OD=14 mm OS=13.5 mm.

Gonioscopy: OU angle is closed by mesodermal tissue. Goniodysgenesis grade III.

Ophthalmoscopy: could not be performed due to corneal edema and pupillary rigidity.

A prenatal ultrasound of a male fetus from a 38-year-old woman with a complicated obstetric history revealed an isolated enlargement of the left orbit and eyeball. Following delivery at 37 weeks of gestation, the infant was referred to the clinic of the Tashkent Pediatric Medical Institute for evaluation due to suspected congenital glaucoma in the left eye.

The child was admitted to the TashPMI clinic on the 16th day after birth, where the diagnosis of advanced stage, uncompensated primary congenital glaucoma (OS) was confirmed. Antiglaucoma surgery was performed on the left eye on postnatal day 20.

Local status. The eyeball has a spherical morphology and increased dimensions. The cornea shows increased diameter and edema. The limbus is dilated. The anterior chamber is deep and the fluid is clear. The pupil is central, round and black. Response to light is minimal. The iris has a smooth pattern and relief. The lens is transparent. The fundus has a diminished pink reflex. Digital tension = 1.0.

The eyeball is spherical. Cornea shows transparency. The anterior chamber is moderately sized and the aqueous humor is clear. The pupil is central, round and black. The reaction to light is preserved. Iris pattern and relief remain constant. The lens is transparent. The fundus reflex is pink. Digital tone is normal.

Tonometry and tonography data.

P t= 19 mm Hg / 28 mm Hg

R o = 11.9 / 11.68

C=0.34/0.09

F = 1.04/1.4

BK(Baker's coefficient)=21.9/136.9

APA (antero-posterior axis) OD=24.2 mm OS=23.5 mm.

Keratometry: OD=13.5 mm OS=10 mm.

Gonioscopy: OD = angle is open. All recognition zones are visible. OS – angle is narrow. Ciliary body is not visible. Goniodysgenesis grade II. Ophthalmoscopy: OD – physiological excavation 10-20 %, OS – excavation 40 %.

Conclusions. Screening ultrasound in pregnant women enhances its capabilities, particularly in visualizing fetal eye conditions for early diagnosis of suspected glaucoma. This visualization serves as a cost-effective and safe screening technique, providing essential data for ophthalmologists. Eye development is readily observable and diagnosable beginning in the second trimester, especially after 17 weeks of gestation. While ultrasound provides valuable insight into fetal eye conditions, definitive diagnosis and confirmation of congenital glaucoma typically requires more specialized postnatal ophthalmic examinations.

Prospects for further research: The findings suggest the need for additional ultrasound screening of pregnant women to facilitate early detection of congenital glaucoma in newborns.

Funding. This study did not receive funding.

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УЗД ЯК ІНСТРУМЕНТ СКРИНІНГУ ГЛАУКОМИ ПЛОДА: КЛІНІЧНИЙ ПІДХІД

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Резюме.

Ініціатива Всесвітньої організації охорони здоров'я VISION 2020 визнала дитячу глаукому третьою провідною причиною значних порушень зору і сліпоти у дітей в країнах з низьким і середнім рівнем доходу, на яку припадає 5 % світової дитячої сліпоти. Дитяча глаукома є критичним станом в дитячій офтальмології, оскільки спричиняє пошкодження зорового нерва та гангліозних клітин.

Мета дослідження – оцінити доцільність проведення ультразвукового скринінгу у вагітних жінок для раннього виявлення ймовірної вродженої глаукоми у плода.

Матеріал і методи дослідження. Для цього було проведено комплексне ультразвукове обстеження 500 вагітних жінок на базі Республіканського центру скринінгу матері та дитини у 2022-2024 роках. Статтю підготовлено на основі результатів запланованого дисертаційного дослідження (Шифр реєстрації – B2023.3.DSc/Tib902; строки виконання: 2022-2025 рр.).

ISSN 2226-1230 (PRINT) ISSN 2413-4260 (ONLINE)
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Результати. Ультразвукові дослідження вагітних жінок надають важливу інформацію для офтальмологів. Ультразвуковий скринінг вагітних з моніторингом орбіт та очних яблук плодів проводився з 20 по 40 тиждень вагітності. Із загальної кількості було проаналізовано дані про 2 (0,4 %) випадки патології очей плода, в яких були виявлені ехографічні ознаки глаукоми, відомі як буфтальм. Після народження у дітей було підтверджено підозру на вроджену глаукому, що потребувало екстреної госпіталізації та хірургічного втручання. Це особливо важливо для дітей з вродженою глаукомою, щоб запобігти сліпоті та порушенням зору в майбутньому.

Висновки: Ця візуалізація може слугувати економічно ефективним і безпечним методом скринінгу, який надає важливу інформацію для офтальмологів. Комплексна ультразвукова оцінка ембріональних очних структур може надати важливу інформацію та виявити вроджену глаукому у немовлят. Точна діагностика та верифікація вродженої глаукоми, як правило, вимагає більш спеціалізованого офтальмологічного обстеження постнатально.

Ключові слова: вроджена глаукома; ультразвукове дослідження плода; скринінг.

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Received for editorial office on 11/06/2024
Signed for printing on 15/09/2024