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CLINICAL CASES OF ANALYZING OBSERVATIONS OF COMBINED CONGENITAL MALFORMATIONS OF THE BRAIN AND VISION IN NEWBORNS

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Abstract:
The present study is aimed at investigating clinical and diagnostic features of combined congenital malformations of the brain and vision in children.

Keywords: Congenital Malformations, Brain, Vision, Children , Clinical Cases.

INTRODUCTION. The relevance of this study is directly related to its potential impact on public health, the quality of life of children and their families, and the development of scientific knowledge in pediatrics, neurology, and ophthalmology [1, 3, 7]. Congenital malformations of the brain and vision represent a serious problem in medical practice, requiring a comprehensive approach to diagnosis and treatment [2, 6, 12]. They can significantly limit a child's abilities and affect his/her physical, cognitive and social development. Early detection and adequate treatment of these malformations can significantly improve the prognosis and quality of life of children [4, 14, 16]. However, despite significant advances in medicine, early diagnosis and treatment of congenital malformations of the brain and vision remain a challenge. Many of these malformations may manifest a variety of clinical symptoms, which makes their timely detection difficult. Moreover, it is not always possible to determine the exact cause of their occurrence [5, 11, 13]. Thus, research aimed at studying the clinical and diagnostic features and risk factors of congenital brain and vision malformations is highly relevant to improve early diagnosis, effective treatment, and prevention of complications [8, 15]. This study is also relevant to the advancement of scientific knowledge in the field of medicine. Understanding the molecular, genetic, and environmental factors that influence the development of congenital brain and vision malformations will help to develop new diagnostic and treatment methods and identify potential targets for the development of new drugs and technologies [9, 17]. Thus, this study is highly relevant from practical and scientific points of view, representing an important contribution to the development of medical science and practice [10].

PURPOSE OF THE STUDY: to investigate clinical and diagnostic aspects of combined congenital malformations of the brain and vision in children with determination of perinatal risk factors for the development of these malformations, peculiarities of clinical manifestations depending on the form of brain malformation and neuroimaging parameters of the study.

MATERIALS AND METHODS OF THE STUDY: As materials of the study we used the data obtained from children aged from birth to 1 year, in whom congenital malformations and anomalies of vision development, combined with structural changes of the brain, and manifestations of complications in the form of epileptic and non-epileptic seizures were detected. The study was conducted on the basis of the Departments of Ophthalmology, Pediatric Neurology and Pediatric Neurosurgery of MK SamSMU (Multidisciplinary Clinic of Samarkand State Medical University), for the period 2021-2024. The total number of patients included in the study is 27 children, who constituted the main group of patients; for comparative analysis, a control group consisting of 20 healthy children of identical age and sex was used. Medical records, results of clinical examinations, neuroimaging data (magnetic resonance imaging of the brain), results of electroencephalography and other laboratory and instrumental research methods (used in ophthalmology) were used to collect data on patients. Data analysis was performed using statistical methods such as comparative analysis, correlation analysis, using specialized software for statistical processing and data visualization. The described materials and methods will allow us to conduct a comprehensive study of clinical and diagnostic features of congenital malformations of the brain and vision in children, as well as to evaluate the effectiveness of various diagnostic methods.

RESULTS OF THE STUDY. The analysis of perinatal risk factors and clinical and neurological signs has provided valuable information on the impact of different

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conditions of pregnancy and childbirth on the health of children, as well as on specific manifestations of neurological conditions depending on perinatal conditions. Numerous studies have pointed to a number of perinatal factors that may have a negative impact on neonatal brain and visual development. Some of these factors may include preterm labor, low birth weight, hypoxia, infections during pregnancy, fetal circulatory disorders, and factors associated with placental malformation. Studies also show that perinatal risk factors may vary according to socioeconomic conditions. Analyzing the results of the study allows us to see that in the control group, the mothers at the time of delivery were between 20 and 35 years of age (85%), at the same time the mothers in the main group, tended to be older than 35 years of age, which amounted to 73%, where the reliability varied within $p<0.001$. The study of the level of somatic status, also indicates the difference in the comparative groups, so in the control group women were healthy in 89% of cases, at the same time women from the main group were characterized by a diverse pathological background of somatic changes, more often in the aggregate of several problems, with the indicators of anemia in this group corresponded (varying degrees) to 100%; the occurrence of specific bacterial and viral infections (TORCH-infection) 69% of cases; problems in the cardiovascular system in 10% of cases; the level of impairment of the cardiovascular system in 10% of cases. The fact of late or insufficient treatment in the polyclinic (primary care) of women of the main group before and during pregnancy, absence or insufficient intake of prophylactic drugs such as folic acid, vitamins and minerals, iodomarin, and inadequate nutrition of women due to their reduced social status was not insignificant. It is known that the nature of the course of pregnancy, plays a leading role in the development of a healthy child, therefore, the presence of all pathological signs at all stages of pregnancy remains important. It was noted that in the women of the main group, pregnancy proceeded with toxicosis (in different terms); the threat of termination of pregnancy was noted in 34% of cases in the first trimester and 29% in the third trimester of pregnancy, with diabetes (gestational) detected only at the end of the third trimester in 5% of cases, and pyelonephritis (gestational) in 16% of cases, where the reliability corresponds to $p<0.005$, compared to the control group. Literature scientific sources, confirm the evidence of the relationship between the frequency of pregnancy of women with the formation of the child's body, as a result of the frequency of pregnancy in the control group (healthy children) in 56% and in the main group in 33% of cases - these births were the first births, where the reliability is equal to $p>0.05$; in some cases, there is a lack of compliance with the interval between pregnancies (in some cases the interval is from 9 months to 1 year), as a factor of depletion of the reserve capabilities of the body of women. In addition, the outcome of previous pregnancies is important in the birth of children with pathological abnormalities, which can be traced in Table 1.

Table 1
History of previous pregnancy outcomes in the comparison groups

Signs of control	Control group (20)	Main group (n=29)
No pathology	93,1	24
Undeveloped pregnancy, spontaneous miscarriages	3,0	36,7
abortions	3,0	5,5
Ectopic pregnancies	0	3,7
Gynecologic infection (history)	3,0	13,9
Stillbirths	0	6,11
Birth of a premature baby	3,0	25,9

The birth history of the children in the comparative groups was as follows: mothers in the main group had obstetrical disorders, such as fetal malposition; weakness of labor, which required labor stimulation (33%) or operative delivery (42.52%). Independent labor was noted in 26.66% of cases in the main group, whereas in the control group more than 80% of cases ($p<0.01$). When considered separately in women of the control group, 12% of cesarean sections were performed in emergency, while in planned (in the same group) only 3.0% of cases. In the comparison group, emergency cesarean section was performed in 35.64% of cases and elective cesarean section in 6.01% of cases, respectively ($p<0.05$). At the same time, clinical and neurological signs vary depending on the predisposing factors (pre- and perinatal disorders),

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reflecting on the stages and forms of the disease. Children with cerebral malformations show various neurologic symptoms such as delayed psychomotor development.

Signs of developmental control, epileptic seizures; these signs may manifest with varying degrees of severity depending on the specific malformation, its location and its impact on brain function. It should be noted that, structural lesions (congenital) of the brain, have in many cases combined lesions: multiple stigmas of embryogenesis, heart defects, abnormalities of development of internal organs, hearing impairment, visual impairment. The studied analysis of pre and perinatal risk factors, peculiarities of clinical and neurological signs in children with brain malformations is an important aspect in the diagnosis and further therapy of this category of children. On the example of a clinical case, all the features of combined brain and vision malformations are described in detail.

Clinical case. Patient S., a boy, was born in 2021, from 1 pregnancy, born on the background of complete placenta previa, delivery at 30 weeks, by cesarean section on the background of placental abruption, Apgar scale 3-5 points, birth weight 1610 g, height 37 cm. The child was in intensive care, 15 days of ventilator, with further transfer to the neonatal pathology department (children's multidisciplinary clinic in Samarkand). In anamnesis on the 7th-8th day from birth, the child had convulsive seizures. The patient's mother was 33 years old at the time of delivery, cytomegaloviruses were detected in the blood, the parents' marriage was unrelated, heredity was not aggravated, during pregnancy, the woman started taking folic acid and vitamins only at late terms. At the time of examination 2023 (the patient is 3 years old), in the dynamics of the formation of hydrocephalus, porencephalic cyst (MRI brain): hypoxic-hemorrhagic lesion of the brain substance, hydrocephalus, cystic-degenerative changes in the brain substance of the right hemisphere. EEG: epileptiform activity in the temporal region, slightly more on the left than on the right, in the occipital parietal region with accent on the left, multifocal with inconstant spread to the same hemisphere and with a tendency to diffuse spread, flash-oppression patterns are observed. Brain MRI for 2021 (after birth). Internal multilevel hydrocephalus occlusive hydrocephalus, isolated BW, adhesions in the 3rd ventricle, suspected occlusion of the vas deferens. Diffuse leukomalacia of the cerebral hemispheres with the formation of porencephalic defects, CNS lesion. Diagnosis at the age of 2 months (after discharge from DMC of Samarkand): Obstructive progressive posthemorrhagic post inflammatory internal tetraventricular hydrocephalus. Tetraparesis. OU Retinopathy of prematurity, active phase, stage III. Epileptiform activity with high index representation during wakefulness and sleep, modified hypsarrhythmia, epileptic spasms were also recorded. TBS radiography on 30.05.23 - radiologic signs of subluxation of both TBS. Daily VEM dated 13.04.2023: Stable interhemispheric asymmetry of cortical rhythmics of wakefulness and sleep due to decreased brain bioelectrical activity and suppression of physiologic patterns in the right hemisphere is registered. The cortical rhythm of wakefulness and sleep in the left hemisphere is generally formed satisfactorily, an increased index of diffuse beta-activity is noted. In wakefulness and sleep, a stable regional delta-delay in the right frontal-central region is registered, with rare spikes of epileptiform activity in the structure of the delta-delay in sleep. Regional epileptiform activity was registered in sleep: - in the right central-vertex region - in the left frontal region, with involvement of vertex leads. Movement disorders syndrome: spastic tetraparesis. Delayed speech and cognitive development. Structural focal epilepsy, medical remission since 03.2022. The patient constantly takes: Valproic acid in drops 13 drops 3 times a day in 18 hours; EEG 08.09.2023. Asymmetry of cortical rhythmicity due to suppression of b.e.a. under the electrodes of the right hemisphere. In the left hemisphere the main activity is formed within the age norm. Increased index of beta oscillations. In wakefulness pathological activity is registered: - regional epileptiform activity in the right temporo-central-frontal area in the form of single sharp waves, spike waves in the structure of continued regional slowing down in the same area, amplitude up to 20 μV - regional continued slowing down of delta-, theta-band in the eve posterior temporo-viso-central-frontal area. Ophthalmologist 06/09/2023. Background Retinopathy and retinal vascular changes Atrophy of the optic nerve. Disease of visual conducting pathways unspecified.

Conclusion: OI - pathology of conductive pathways, ChASN, dystonic retinal angiopathy. 12.09.2023 Complaints: does not walk independently, does not stand, delayed motor skills. The head is hydrocephalic. OG 54.5 cm. OHC 51 cm. Cranial nerves: 2 (Opicus) pair - visual concentration is present, gaze does not fixate, does not trace. 7 (Faciais) pair - eye slits S=D, face symmetrical. 8 (Acusicus) pair: hearing is tentatively unimpaired, localizes sound. 12 (Hypogossus) pair: tongue in the oral cavity along the midline. Reflex-motor sphere: movements in joints: limitation of supination of the left forearm and hand. Limitation of extension in, Hamstring syndrome. Graciis-syndrome is positive on both sides. Muscle tone is increased by spastic type,

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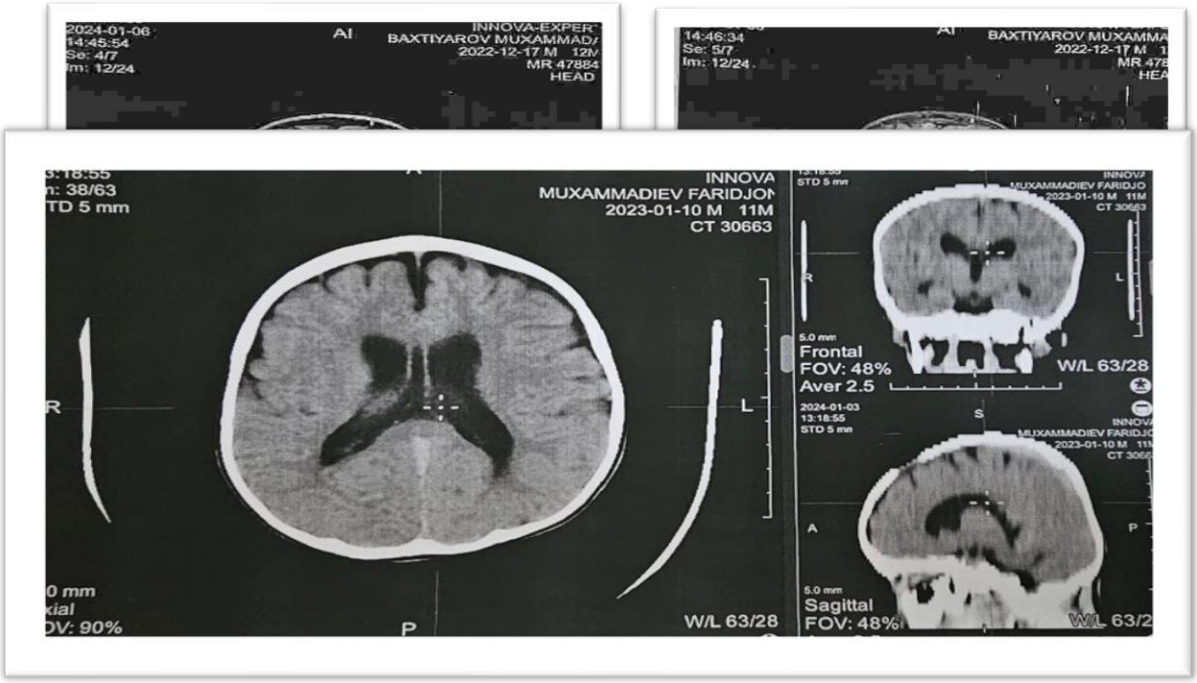
D≤S. Tendon



the girl

reflexes: animated with expansion of reflexogenic zones, S≥D. Abdominal reflexes are alive, symmetrical. Babinski's symptom on both sides. Motor skills: Head position is not changed, head retention is difficult, holds for 10 seconds. Turns on the side + nipples - rarely on the stomach. Does not sit, posture does not hold. Does not get on all fours. In the position on the stomach can turn his head to the sides, demonstrate short-term support on the forearms. In passive verticalization, there is support on legs, makes stepping movements with hip translation and crossing in the lower third of the shins, flat-valgus setting of the feet, not to the extension of the knee joints. Mental status: Speech - up to 5 babbling words. Reacts to intonation and voice, laughs, follows simple instructions - give hand, show nose. Pelvic functions - does not control. In behavior is emotional, demonstrative.

Clinical case. Patient M, a girl, born in 2022, from 3 pregnancies, 3 births (twins, the second child stillborn), gestational age 34-36 weeks, the child was born weighing 2020g, Apgar scale 5-6 points, emergency cesarean section was performed. The mother had BP 160/100, edema (diagnosis of Preeclampsia 2 degree), the parents were related (cousins). After delivery, the child was transferred to the intensive care unit, on the SIPAP machine. In the dynamics of observation in 2024, has a decrease in the volume of the skull from the norm, does not fix her gaze, does not hold her head, does not sit independently, in speech activity a few separate syllables are noted. According to her parents, periodic vomiting, screaming and crying are frequent complaints. MRI revealed the following signs: MR tomography using T1, T2 modes in sagittal, transversal and coronal projections. Internal multilevel hydrocephalus occlusive, isolated lateral ventricles, adhesive changes in the third ventricle. Diffuse leukomalacia of the cerebral hemispheres with the formation of porencephalic defects, enlargement of the subarachnoid space in the frontal temporal-parietal regions on both sides. The corpus callosum is thinned. Retinal detachment on both sides. At the same time, blood biochemistry analysis of the mother at the time of delivery, for Homocysteine -15.2 mmol/L; F T-3 - Triiodothyronine free -8.2 pmol/L; Folat III - Folic acid - 4 ng/mL. Literature scientific sources confirm that the risk of formation of congenital brain malformations, depend on the effect on folic acid metabolism, reducing its level in the serum of a woman, thereby causing hyperhomocysteinemia. Homocysteine, in turn, has a pronounced cytotoxic effect, including on the cells of the growing embryo, thereby causing fetal malformations (Maychuk V.O. 2013).

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Fig. (1,2,3) MRI of patients with congenital combined brain and vision malformations.

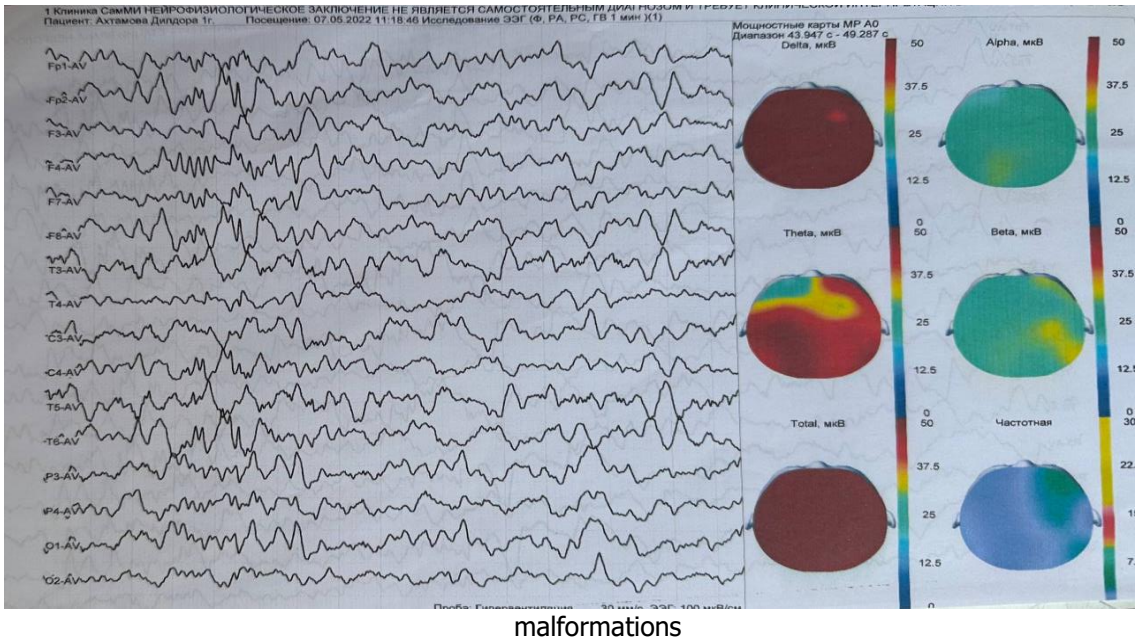

Fig. 4 EEG of patients with congenital combined brain and eye malformations

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of the brain and eyes

CONCLUSIONS: Thus, a study aimed at investigating clinical and neurological signs and neuroimaging changes in children with congenital malformations of the brain and vision represents a significant contribution to the understanding of these conditions and the definition of approaches to their diagnosis and treatment. From the analysis of perinatal risk factors, it has become clear that certain aspects of pregnancy and childbirth may influence the development of congenital brain and visual malformations in children. This emphasizes the importance of monitoring women during pregnancy and timely diagnosis of possible complications. The study revealed significant associations between clinical manifestations and neuroimaging findings, in children with congenital brain and vision malformations indicates the presence of changes in anatomy, which may reflect the degree of disturbance of normal development. Electroencephalography (EEG) monitoring also play an important role in the diagnosis of paroxysmal disorders in these children. The correlation between clinical manifestations and neuroimaging data emphasizes the need for a comprehensive approach to patient evaluation. In conclusion, it should be noted that the subsequent development of an algorithm of practical and preventive recommendations to improve early diagnosis and treatment of congenital brain and vision malformations in children is an important step in improving the quality of medical care for this category of patients. Careful observation of the patients' condition and regular comprehensive examination can help in timely detection and correction of possible pathologies.

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