Ophthalmological Features of Optic Disc Malformation in Children

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ABSTRACT

Purpose of the study to study the diagnostic criteria for ONH in children

Keywords:

Actuality.

Detection and adequate verification of anomalies of the optic nerve head in children in the first months of life is important not only for their timely ophthalmic rehabilitation, but also plays an extremely important role for genetic counseling and early diagnosis of systemic diseases associated with malformations of the optic nerve in infants [1,2,3,11]. Optic nerve hypoplasia (ONH) occurs in economically developed countries with a frequency of at least 7 cases per 100,000 population and is the cause of low vision and blindness in 5-6% of cases [4,5,6,12].

Due to the small number of publications devoted to ONH and its association with systemic pathology, ophthalmologists are not sufficiently informed about the nature of the course and clinical features of some diseases from this group [7,8,9,10], which leads to a high frequency of diagnostic errors and unsatisfactory functional results of treatment.

Purpose of the study.

To study the diagnostic criteria for ONH in children.

Materials and research methods.

When performing this work, we analyzed the results of neuro-ophthalmological examination and observation of 25 children with ONH aged 7 to 16 years. The material was collected at the Republican Clinical Ophthalmological Hospital. All patients underwent: collection of anamnestic data, clinical and ophthalmological examination.

Research results.

The study found that the main causes of ONH development are perinatal lesions of the periventricular white matter of the fetus, caused by hypoxia-ischemic disorders (32%), intrauterine infection (8%) and chronic toxic effects (4%). ONH can affect both one (44%) and two (56%) eyes with approximately equal frequency. ONH is characterized by typical

ophthalmoscopic manifestations of a decrease in the size of the optic disc (in 100%) of cases, its decoloration (48%), a "double ring" symptom (44%), a corkscrew-like tortuosity of the retinal vessels (32%), and the absence of macular and foveolar reflexes (96%). ONH in all cases was verified in the study of the fundus. Children with ONH have a high level of ametropia (92%), oculomotor disorders (80%), concomitant changes in the anterior and posterior segment (64%), which make it difficult to diagnose and aggravate visual deprivation, which leads to the frequent development of amblyopia.

As a result of neuroradiological studies, pathological changes in the brain were established in 72% of patients. Some of these anomalies may not cause neuroendocrine disorders, in particular, agenesis of the septum pellucidum (found in 32% of patients) and hypo- or agenesis of the corpus callosum (in 40%). In children with bilateral lesions, neuroradiological examination reveals periventricular white matter pathology almost 3 times more often ($p < 0.001$) than in children with unilateral ONH (92.8% and 36.4% of cases, respectively). This indicates that bilateral ONH develops against the background of more severe periventricular white matter lesions and can be considered an indicator of multiple periventricular white matter malformations. If a bilateral anomaly of the optic nerve is found in an infant, a neuroradiological examination and EEG recording should be performed.

All children with ONH showed a decrease in the vertical and/or horizontal diameter of the ONH disk, a decrease in reflectivity and thinning ($p<0.01$) of RNFL in all or (in segmental forms of ONH) in certain quadrants, a decrease in the total average thickness of RNFL (on average, up to 52.1±23.8 μm) compared with the age norm ($p<0.01$). Pathology of the central nervous system is determined by radiological studies in 68% of patients with ONH. Endocrine, neurological and visceral diseases were found in 52% of children with ONH.

Analyzing the data, with ONH, light perception for blue and green colors and the degree of light perception are more affected, which indicates ongoing irreversible processes in the optic nerve and persistent ischemia and reflects the coefficient of vision loss. The obtained data of EEG can serve as a differential diagnostic criterion.

Conclusions.

The main reasons for the formation of ONH are pre- and perinatal lesions of the periventricular white matter of the fetal brain, caused by hypoxic-ischemic disorders (32%), intrauterine infection (8%) and chronic toxic effects (4%).

Due to the prevalence of systemic pathology in children with ONH, when observing them, it is necessary to use methods of radiation diagnostics: in all cases - NSG, and in children at risk (with bilateral lesions, with a combination of ONH with neonatal hypoglycemia and / or prolonged jaundice) - MRI and ultrasound examination of the abdominal cavity and retroperitoneal space.

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