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AN INTEGRATED APPROACH TO THE DIAGNOSIS OF OPTIC NERVE ABNORMALITIES: FROM CLINICAL OBSERVATIONS TO INNOVATIVE TECHNIQUES (REVIEW ARTICLE)

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Topicality.

In recent decades, the attention of specialists has been increasingly focused on the problem of abnormalities in the development of the optic nerve [1, 7, 14], which is due to their significant impact on visual function and quality of life of patients. Developmental abnormalities of the optic nerve, including optic disc coloboma, cavernous anomaly, and others, are complex and diverse in nature. These can range from minor abnormalities with little to no effect on vision to serious conditions leading to significant visual impairment or even blindness.

The study of these anomalies requires a comprehensive approach, including a detailed clinical examination, the use of modern diagnostic technologies and the interaction of narrow specialists [2, 3]. Early diagnosis and accurate classification of abnormalities are very important for the development of effective treatment and rehabilitation strategies.

Among the key diagnostic criteria for abnormalities in the development of the optic nerve are the features of the structure of the optic disc, the presence of concomitant ophthalmic and systemic disorders, as well as genetic factors [4]. Advances in genetic research and neuroimaging are contributing to a better understanding of the etiology and pathogenesis of these conditions, which opens up new prospects for their correction and preventing disease progression.

The relevance of the topic is due not only to medical, but also to social aspects, since visual impairments significantly affect the adaptation of individuals in society, their educational and professional opportunities. In this regard, improving the efficiency of diagnostics and developing new approaches to the treatment and rehabilitation of patients with optic nerve anomalies are becoming priority areas in modern ophthalmology.

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Discussion.

Congenital and hereditary abnormalities of the optic nerve highlight their role in the development of visual dysfunction. A wide range of optic nerve abnormalities are still under-covered, including coloboma, hypoplasia, hereditary optic neuropathy, Aicardi syndrome, optic nerve drusen, optic nerve fossa, and "morning glow" syndrome. The attention of researchers should be focused on abnormalities affecting the visual system and associations with neurological and systemic pathologies and, most importantly, the correct identification of optic nerve disease [5, 7].

A study by Skriapa M.A. et al. (2018) analyzed the prevalence, clinical characteristics, and associated morbidity in children with optic disc coloboma (PCD). The study was population-oriented, cross-sectional, involving 31 children aged 2 to 18 years diagnosed with KZN. It is part of a wider cohort of 184 children with congenital optic disc malformations. The prevalence of KZN was 8.9 per 100,000 children. Of the 31 patients, 18 had unilateral CKN. The best corrected visual acuity (SDA) in eyes with CKN ranged from blindness to 1.3 (median 0.3). LSVO was 0.82 in eyes with isolated CNN (range 0.4–1.3) and 0.15 in eyes with concomitant macular involvement (range 0–0.5). Nystagmus was observed more frequently in patients with bilateral CNN (9 of 13 versus 3 of 17). Two patients were found to have retinal detachment.

The study found that optic disc coloboma is the second most common optic disc abnormality after optic nerve hypoplasia in this cohort. The children exhibited a wide range of ocular comorbidities. Isolated NCN without macular involvement was not associated with profound vision loss, highlighting the importance of detailed diagnosis and an individualized approach to the treatment and rehabilitation of patients with such diseases [14].

Martin L.L. (2022) studied the case of bilateral coloboma of the optic disc associated with the absence of the corpus callosum. This is a rare pathology that highlights the importance of evaluating systemic abnormalities in the diagnosis of optic disc coloboma. The author discusses that the optic disc coloboma is a well-defined excavation in the optic nerve head, usually located below with the neuroretinal rim intact at the top. The theory of the development of coloboma of the optic disc, which has evolved over time, is considered.

The author's research emphasizes the need for a systematic assessment in the diagnosis of optic disc coloboma, given the potential association with serious systemic abnormalities such as the absence of a corpus callosum. Awareness of such an association is important for ophthalmologists and pediatricians to provide a comprehensive approach to the management and treatment of patients with such conditions [12].

Hwang I. et al. (2021) studied maculopathies associated with Morning Glow Disc Abnormality (ADUS) in their papers, revealing the role of multimodal imaging in the assessment of this rare condition. ADUS is a rare congenital condition, most commonly found in white women during childhood, that reduces vision. About half of the cases go on to develop maculopathy or retinal detachment of the posterior pole.

The use of multicolor and optical coherence tomography makes it possible to identify the morphological characteristics of the optical disc and macula.

The authors hypothesize that the centripetal internal gravity of the retina and fluctuations in cerebrospinal fluid pressure play an important role in the accumulation of internal retinal fluid in the pathology of retinoschisis in ADUS. Further research may shed light on a potential causal relationship between ADUS and retinoschisis.

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The study highlights the importance of multimodal imaging in the diagnosis and evaluation of maculopathy associated with ADUS and suggests further investigation into the internal mechanisms of this disease. This case makes an important contribution to the understanding of the clinical picture and potential pathways of maculopathy in ADUS, expanding knowledge of the pathophysiology and treatment options for this condition [8].

Study by Kim J.A. et al. (2019) focuses on the study of the development of optic disc torsion in Korean children. The authors analyze sequential photographs of the optic disc taken at least one year apart for 173 eyes in 173 Korean children. The purpose of the study is to document the development of disc torsion.

The axis angle of the vertical disc in each hazelnut image was measured, with the axis of the center of the disc-fovea set to 0°. Associated changes in optic disc morphology were assessed by measuring the ratio of horizontal and vertical disc diameters, as well as the ratio of the maximum width of parapapillary atrophy to the vertical diameter of the disc.

The authors discuss the possible mechanisms of the development of optic disc torsion in children and the relationship of this condition to myopia and other eye diseases. Particular attention is paid to the effect of disc torsion on visual function and the need for further research to better understand this phenomenon.

The study highlights the importance of monitoring optic disc torsion in children, especially among those at risk of developing myopia and other comorbidities. The findings may contribute to the development of strategies for early diagnosis and prevention of potential visual impairment associated with disc torsion [10].

Amador-Patarroyo M.J. et al. (2015) view congenital optic nerve anomalies as a group of structural malformations of the optic nerve and surrounding tissues that can cause congenital visual impairment and blindness. The authors emphasize that each abnormality in this group becomes more prevalent as our ability to differentiate them improves through better case characterization and access to improved medical technology such as neuroimaging and genetic analysis [3].

Improvements in medical technology, such as neuroimaging and genetic analysis, have expanded our understanding of these abnormalities. Visual impairment may not be the only problem in these patients; Some abnormalities may be due to ophthalmologic, neurological, and systemic features that will help the physician identify and predict possible outcomes in these patients, sometimes potentially life-threatening

Study by Liu T.Y.A. et al. (2021) presents an innovative approach to the detection of visual disc abnormalities in color fundus photographs using artificial intelligence. The authors developed and evaluated an artificial intelligence model capable of automatically identifying various optic disc abnormalities, which represents significant progress in the field of ophthalmic diagnosis.

The developed model demonstrates high accuracy and sensitivity in detecting anomalies, which is confirmed by comparison with the traditional assessment by specialists.

The authors discuss the potential of using artificial intelligence and machine learning to improve screening and early diagnosis of eye diseases, especially in settings with limited access to eye care.

The study highlights the significant potential of applying deep learning in ophthalmology to improve the diagnosis and monitoring of optic disc abnormalities. The development and implementation of such technologies can significantly improve the quality and accessibility of eye care, especially in under-resourced settings [11].

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This research makes an important contribution to the development of artificial intelligence technologies in medicine, opening up new horizons for the diagnosis and treatment of ophthalmic diseases

Study by Ceylan O.M. et al. (2021) presents a unique case study of a patient with multiple optic disc fossa for 21 years. Optic disc fossae, a rare birth defect usually manifesting as ovoid, gray-white notches in the lamin cribrosis of the optic disc. The incidence is 1 case per 11,000 population, affecting both sexes equally, predominantly unilaterally in 85-90% of cases and bilaterally in 10-15% of cases.

Serous macular detachment is estimated to affect 25-75% of patients with NFD.

This study presents a case of dual NNV in the right eye and single NNV in the left eye in a patient who was followed for 21 years due to partially compensated esotropia.

The study highlights the importance of long-term follow-up and monitoring of patients with NWD, given the potential impact of this condition on visual function and the possibility of developing serous macular detachment. The authors point to the need for a better understanding of the mechanisms of development and progression of ophthalmic abnormalities associated with NNF and the development of strategies for their effective management and treatment [6].

Optic nerve aplasia (ONA) is a rare disease in which the optic disc, retinal vessels, and retinal ganglion cells are missing, resulting in lifelong vision loss. It can be one-sided or two-sided. Unilateral OH is usually associated with normal brain development, while bilateral OH may be accompanied by malformations of the central nervous system (CNS).

Based on a multicenter study in which 9 cases of OH were described, the age of the patients (range 10 days to 2 years, median 9 months), sex (6 boys), and bilateral involvement (7 cases) were summarized. The most common ophthalmic disorders were glaucoma, microroots, persistent pupillary membrane, iris coloboma, aniridia, retinal dysplasia, retinal atrophy, choroidal coloboma, and persistent fetal vasculature. Systemic abnormalities included facial dysmorphism, cardiac, genitourinary, skeletal, and developmental defects.

The study also identified a mutation in the BCOR gene in one case, highlighting the genetic nature of some cases of OHA. Neuroimaging was performed for all patients, showing the absence of an optic nerve and, in bilateral cases, the absence of chiasm. One case showed the presence of vestigial optic nerves and chiasm despite the absence of a visible optical disc on ophthalmologic examination. This suggests that OHA may be an extreme form of optic nerve hypoplasia, the severity of which may be related to the timing and location of the embryological defect.

It can manifest as an isolated disease or in association with other congenital anomalies. It is important to pay attention to the possibility of systemic disorders, especially in bilateral OA. The presence of an eyeball undoubtedly indicates that an optical trunk was present at some point, although this trunk may not have been filled with axons. The proposed theory that aplasia may be the result of abnormal vascularization secondary to defective germinal cleft formation is supported by the presence of retinal coloboma in some cases, as well as in some cases of severe optic nerve hypoplasia [13].

The prognosis for patients with optic nerve abnormalities varies and depends on the specific type of abnormality, its extent, and the timeliness of treatment. In some cases, surgery is possible, such as retinal detachment associated with "Morning Glory" syndrome [9].

Early diagnosis of optic nerve abnormalities is critical to prevent the progression of the disease and reduce the risk of further complications, including vision loss.

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Effective management of optic nerve abnormalities requires a comprehensive approach that includes ophthalmology, neurosurgery, and genetic counseling, especially in cases where the abnormalities are associated with systemic diseases.

Based on the review of the article, it can be concluded that abnormalities in the development of the optic nerve are a complex group of conditions that require an individualized approach to diagnosis and treatment. Effective management of these conditions depends on a number of factors, including early diagnosis, a multidisciplinary approach to treatment, and, in some cases, surgery. Further research is needed to improve understanding of the etiology and pathogenesis of these abnormalities, as well as to develop more effective treatment and rehabilitation strategies for patients

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