Leber’s hereditary optic neuropathy: the peculiarities of inheritance and genetic consultation

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Introduction

The study is focuses on Leber hereditary optic neuropathy (LHON), a mitochondrial disease characterized by bilateral loss of vision and early onset (age 20-35 years).

Aim of the study

To analyze current scientific data about Leber’s hereditary optic neuropathy and the therapeutic strategies in the treatment of patients with LHON.

Materials and methods

In the course of the work, various sources of data were analyzed, including relevant data on the problem in the Republic of Belarus.

Results

The disease occurs all over the world, mostly affecting males (the probability of manifestation is 4-5 times higher).

The prevalence of the disease is 1: 25,000 in the north-east of England, in Russia it is 3.2: 100,000, and it causes 2% of all cases of blindness in Australia. In Belarus, two cases of Leber's atrophy of the optic nerves have been revealed.

The most common cause of LHON is a mutation in the ND-4 gene in the position 11778G>A (replacement of adenine with guanine), which encodes the structure of the NADH-ubiquinone oxidoreductase enzyme. A necessary condition for the phenotypic manifestation of the disease is the presence of a mutation in the state of homo- or heteroplasmy. Further development of the disease involves genes localized in the X chromosome, in this regard, there is unequal occurrence of the disease in men and women. In addition, triggers of LHON may include environmental factors: infection, starvation, hypothermia, stress, general anesthesia. All of the above factors lead to a decrease in the synthesis of ATP, an increase in the number of free radicals and a disturbance of redox processes. Therefore, apoptosis of retinal ganglion cells is subsequently triggered, which leads to degeneration of the optic nerve cells and directly to loss of vision.

Conclusion

A wide variety and similarity of the clinical symptoms of mitochondrial diseases and diseases having different etiology requires a differentiated and competent approach to the diagnosis of patients with Leber's disease.

In the genetic counseling of families, a number of factors must be considered: the presence of a mutation in the carrier in a homo- or heteroplasmic state; incomplete penetrance of the mutation and the relationship between the degree of manifestation of the disease and sex; the impact of some provoking factors on the patient.

The findings of this research are a prerequisite for improving the quality of genetic counseling and for developing new therapeutic strategies in the treatment of patients with LHON.