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A MODERN METHOD IN THE TREATMENT OF MITOCHONDRIAL MYOPATHIES - GENE THERAPY

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A B S T R A C T

In this article, it became known that every healthy person has a 35% chance of having mitochondrial mutations. In order for it to appear, more than 60% of the mitochondria in the cells must have this defect. With our current treatment method, we can save not only patients, but also carriers with gene deletion in their mitochondria, even if the disease does not appear. In particular, in the treatment of myopathies, it is necessary to use not only symptomatic methods, but also genetic engineering methods. Currently, a new method of treating patients with other types of mitochondrial cytopathies is aimed at restoring the structural integrity of mitochondrial genes in the egg cell. With this, we are on the way to improving the health of children who may be born with mitochondrial diseases.

K E Y W O R D S

mitochondrial myopathies, gene mutation, deletion, mutant mitochondria.

Purpose of Work

In the treatment of mitochondrial diseases, including myopathies, it is necessary to use not only symptomatic methods, but also genetic engineering methods. Scientists have identified many genetic mutations that cause mitochondrial diseases. They used this knowledge to create animal models of mitochondrial disease that could be used to test potential treatments. Scientists have also developed genetic tests that allow accurate diagnosis of mitochondrial defects and provide valuable information for family planning. Knowing the genetic mutations that cause mitochondrial diseases opens up the possibility of developing specifically targeted therapies. By better understanding mitochondrial biology, scientists hope to develop unique approaches to treating mitochondrial diseases. Because the cells of people with mitochondrial disease often contain a mixture of healthy and mutant mitochondria, an effective therapy may involve taking over the healthy mitochondria. It may be possible to rescue mutant mitochondria by encouraging them to fuse with healthy mitochondria. Another approach could be to stimulate the birth of new mitochondria, which would help the healthy ones multiply and outnumber the mutants.

The Urgency of the Problem:

Mitochondrial myopathies are a progressive muscle wasting disease that mostly begins in childhood. One in 5,000 people suffer from genetic mitochondrial diseases. Every year, 6,000 to 8,000 children worldwide are born with a mitochondrial disease. Mitochondrial diseases are caused by defects in the mitochondria, the powerhouses found in almost all cells in the body. Mitochondrial diseases that cause serious muscle problems are called mitochondrial myopathies, and mitochondrial diseases that cause muscle and neurological problems are called mitochondrial encephalomyopathies. Currently, our research focuses on the first type of mitochondrial diseases above. A typical human cell relies on hundreds of mitochondria to meet its energy needs. Mitochondrial disease symptoms vary: in most cases, mitochondrial disease is a multisystem disease that affects multiple cells, tissues, or organs. Because muscle and nerve cells have particularly high energy demands, muscle and neurological problems are common features of mitochondrial disease. Other common complications include vision impairment, cardiac arrhythmia (abnormal heartbeat), diabetes, and growth retardation. Usually, a person with mitochondrial disease has two or more of these conditions, some of which regularly coexist and are grouped into syndromes.

The main symptoms of mitochondrial myopathy are muscle fatigue, weakness, and exercise intolerance. The severity of any of these symptoms varies greatly from person to person, even within the same family. In some people, the weakness is most common in the muscles that control the movements of the eyes and eyelids.

Mitochondrial myopathies can also cause weakness and weakness in other muscles of the face and neck, which can lead to difficulty swallowing and, rarely, slurred speech. People with mitochondrial myopathy may also experience muscle weakness in the arms and legs. The degree of exercise intolerance varies greatly between individuals. Some people may only have problems with sports such as running, while others may have difficulty walking, sitting, or standing.

Mitochondrial disease can sometimes cause pain after exercise. Although overexertion should be avoided, moderate exercise can help people with mitochondrial myopathy ease the condition.

Another important aspect of this disease is that this type of disease is treated only symptomatically and not by pathogenetic or etiological methods. The treatment method is also very simple, only aimed at increasing the efficiency of energy metabolism. Another method is aimed at preventing mitochondrial membrane damage using antioxidants. Our treatment below is based on the use of genetic engineering.

Research Methods:

First of all, it is worth mentioning that mitochondrial myopathies are considered a hereditary disease, and its inheritance is passed only from mother to her children. Oxidation process in mitochondria slows down due to mutation, especially deletion of genes in mitochondrial DNA in cytoplasm of egg cell ready for fertilization. As a result, when these mutant mitochondria are given to the cells of the tissue during organogenesis, the work activity decreases and catabolism is disturbed due to the fact that the energy needs of this tissue are not fully satisfied. For example, if mutated mitochondria are transferred to heart muscles, cardiomyocytes are not fully supplied with energy. The only problem with this is the mitochondria defect. Our proposal is to restore the part of mitochondrial genes lost due to deletion. For this purpose, the gene lost due to mutation from the patient's healthy mitochondria and defective mitochondria is artificially isolated and cloned. Amplified genes are combined with vector construct genes. Now we are thinking about using viruses as vectors. We attach the important gene to the genes

in the viral DNA and send it to the sick organism. We theoretically studied the normalization of energy exchange in cells as a result of the recombination of genes delivered to cells by the vector with the lost genes of mitochondria. Currently, a new method of treating patients with other types of mitochondrial cytopathies is aimed at restoring the structural integrity of mitochondrial genes in the egg cell. With this, we aim to improve the health of children who may be born with mitochondrial diseases.

Conclusion:

In conclusion, we should note that every healthy person has a 35% chance of having mitochondrial mutations. In order for it to appear, more than 60% of the mitochondria in the cells must have this defect. With our current treatment, we can save not only patients, but also carriers who have a gene deletion in their mitochondria, even if the disease does not appear.