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Mitochondrial Encephalomyopathy

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Abstract: Mitochondrial encephalomyopathy is a genetic disease caused by mutations in mitochondrial genes (mitochondrial DNA, mtDNA). Different symptoms occur due to mitochondrial or biochemical changes that occur in different tissues. Like many mitochondrial diseases, it occurs in many organs. Representative symptoms include myopathy, encephalopathy, lactic acidosis, and stroke-like episodes. In this article, we will talk about the causes, symptoms, diagnosis and treatment of Mitochondrial encephalomyopathy.

Keywords: Mitochondrial encephalomyopathy, DNA, mutation, MELAS, diabetes, therapy, Wolff-Parkinson, coenzyme.

The purpose of our research: The purpose of our research is to study the causes of mitochondrial encephalomyopathy, its development, diagnosis and treatment methods.

Inspection methods and materials. Mitochondrial DNA (mtDNA) is a 16,569-base circular DNA that encodes 22 tRNAs, 2 rRNAs, and 13 mitochondrial enzyme proteins. Due to mutations in MELAS, an AG mutation at position 3243 and a TC mutation at position 3271 were identified in the tRNA Leu(UUR) coding region. Among them, the most common is the 3243 mutation, which is detected in approximately 80% of patients with MELAS. 3,271 mutations account for about 10%, and there are 3,252 more mutations.

① Short stature

The most common feature of this disease is short stature. Pediatric endocrinologists have tried to use growth hormones to stimulate growth in short patients, but the results have not been successful. It was also ineffective in other patients.

② Muscle disorders

Muscle disorders usually precede the first "seizure-like symptoms." A 4-month-old baby's limbs appear limp and weak, and symptoms of fatigue and weakness appear quickly during exercise. Muscle dysfunction is usually more severe in muscles closer to the body than in muscles farther away. Muscle tissue is usually thin. An increase in the activity of creatine phosphokinase in the blood is also observed. In some patients, it is accompanied by degenerative and reproductive changes. These patients are more likely to develop polymyositis, which is usually more serious than the amount of muscle loss. Meanwhile, problems such as developmental delays, learning difficulties, and difficulty concentrating appear before the first seizure.

③ Seizure-like symptoms (MELAS)

The starting point of this disease is first seizure-like symptoms between 4 and 15 years of age. It is called a "seizure-like symptom" because no vascular changes occur due to inflammation or

atherosclerosis in the brain. These symptoms are called MELAS. This symptom occurs in several members of a family with the same mutation.

Early symptoms include vomiting, headache, convulsions, and visual abnormalities, and rarely, sensory abnormalities, hemiplegia, and aphasia may also occur. Vomiting or headache can last from several hours to several days. These symptoms may manifest as temporary hemiplegia or unilateral visual field defects lasting from days to weeks. Usually, most family members of a MELAS patient complain only of migraines.

After these symptoms appear, a CT (computed tomography) or MRI scan of the brain may be performed to detect a clear pattern similar to an infarct. Although these features may disappear, they may later leave behind sequelae of brain atrophy and calcification. Cytological examination of this area may reveal widespread infarcts in the brain, cerebellum, or brain stroma.

Most patients are normal, but when brain disease develops, it progresses to dementia. Patients may appear apathetic or cachectic.

④ Neurological diseases

Neurologic features include ataxia, tremor, dystonia, visual disturbances, and cortical visual disturbances. Some patients also show partial myoclonus. Facial muscle disturbances or ptosis may occur, and retinal pigmentary degeneration may also occur. Another common feature is hearing loss, which occurs in 25% of patients. Myocardial dysfunction is not a common feature, but occurs in 10% of patients. Usually this is a hypertrophic myocardial disorder. ECG test results are often abnormal.

⑤ Diabetes

MELAS mutations can cause diabetes. Type II diabetes is commonly present and, rarely, insulin-dependent diabetes is also present.

⑥ Histological characteristics

Fatigue of red blood cells (red cells) occurs in the muscles. In this case, an increase in the number and size of mitochondria is visible through an electron microscope. An important feature of this disease is lactic acid. It usually does not cause full-blown acidosis and may not occur in patients with central nervous system involvement. Cerebrospinal fluid may also increase. Blood level is normal. Cerebrospinal fluid protein levels may be moderately elevated.

⑦ As with other Wolff-Parkinson-White syndromes, conduction disturbances occur and electrocardiograms often show abnormal results. Enlarged mitochondria are observed in nerve fibers. In addition, different problems may arise depending on the individual. For example, some patients may have symptoms such as nephrotic syndrome and glomerulosclerosis, while others may have symptoms such as terminal neuritis and ischemic colitis with rhabdomyolysis, which causes muscle breakdown.

Treatment of this disease, like other mitochondrial diseases, uses supportive care.

The use of riboflavin was effective in patients with complex I deficiency and the T-C 3250 mutation. A 2-year-old patient with muscular dystrophy who was unable to climb stairs and had difficulty walking was administered 20 mg twice daily. As a result, the muscles became stronger, and no more deformities were observed after three years of follow-up.

Coenzyme Q10 has been effective for many patients. Muscle wasting was somewhat restored and serum lactic acid levels also decreased. However, it had no significant effect in reducing lactate levels in cerebrospinal fluid. Coenzyme Q has been used in doses ranging from 30 to 90 mg per day. It has been concluded that a higher dose of approximately 300 mg per day is required for optimal results in this disease.

Dichloroacetic acid is an effective agent for reducing lactate levels in serum and cerebrospinal fluid. In addition, arginine is also known to be effective against strokes. Monitoring and treatment of diabetes is also necessary.

Conclusion. In conclusion, the symptoms caused by Mitochondrial inheritance are very diverse. Characteristics of this disorder include normal development before severe, progressive brain damage. These symptoms include: short stature, diabetes, neurological diseases, muscle disorders. This disease is diagnosed on the basis of clinical appearance and genetic examination of mitochondrial DNA.

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