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Perspective

Importance in Mitochondrial Disorders

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DESCRIPTION

Mitochondrial disease or mitochondrial disorders refer to a group of disorders that affect mitochondria, a small compartment found in almost every cell in the body. The main function of mitochondria is energy production. More energy-demanding organs, especially the heart, muscles, and brain, need more mitochondria to produce more energy. When the number or function of mitochondria in a cell is destroyed, less energy is produced, resulting in organ dysfunction. Various symptoms can occur, depending on which cells in the body have destroyed mitochondria. Mitochondrial disease causes a variety of health problems such as malaise, weakness, metabolic stroke, attacks, cardiomyopathy, arrhythmias, developmental or cognitive impairment, diabetes, hearing impairment, vision, growth, liver, gastrointestinal or renal function. There is a possibility. These symptoms can occur at any age, from childhood to late adulthood. Mitochondrial dysfunction occurs when mitochondria are not functioning due to another disease or condition. Many disorders cause secondary mitochondrial dysfunction and can affect other disorders, including:

- Alzheimer's disease.
- Muscular dystrophy.
- Lugeric's disease.
- Diabetes.
- Cancer Types

Types of mitochondrial disorder's

Examples of mitochondrial diseases are:

- Mitochondrial myopathy
- Mitochondrial diabetes
- This combination at an early age may be due to mitochondrial disease
- Diabetes and deafness can be found together for other reasons
- Leber's Hereditary Optic Neuropathy (LHON)
- Loss of vision beginning in young adulthood
- An eye disease characterized by progressive loss of central visual acuity due to degeneration of the optic nerve and retina
- 1 in 50,000 people are affected in Finland
- Leigh's encephalopathy, subacute sclerosing encephalopathy
- After normal onset, the disease usually begins in the second half of the first year of life, but can also occur in adulthood.
- There is a rapid decline in function characterized by seizures, altered state of consciousness, dementia, respiratory arrest
- Neuropathy, Ataxia, Retinitis Pigmentosa And Ptosis (NARP)
- Progressive symptoms described by the acronym.

CAUSES

Mitochondrial genetic damage is caused by changes (mutations) in either mitochondrial DNA or nuclear DNA and can cause mitochondrial dysfunction. Most DNA, a genetic material that is passed down from parent to child, is packaged in the nucleus of each cell (known as nuclear DNA). However, mitochondria (the structure of each cell that produces energy) contain a small amount of unique DNA called mitochondrial DNA.

If the mutated gene that causes the disease is on the X chromosome, which is one of the two sex chromosomes, the disease is considered to be associated with the X chromosome (the Y chromosome is another sex chromosome). Females have two X chromosomes, and males have an X and Y chromosomes. X-linked states can be X-linked dominant or X-linked recessive. Inheritance is X-linked dominant when one copy of the altered gene in each cell is sufficient to cause the condition. A X-linked dominant female has a 50% chance of inheriting the condition to her son or daughter with each pregnancy. A man with X-linked dominant conditions passes the condition to all daughters, not to her son. If a gene on the X chromosome causes the condition in a man with one gene mutation (only one X chromosome) and a woman with two gene mutations (with two X chromosomes), the inheritance is X-linked recessive is Females with X-linked disease convey the mutation to all sons and daughters. This means that all her sons will get sick and all her daughters will be carriers. A man with X-linked recessive disorder passes the mutation to all daughters (carriers), not his son.

Diagnosis of mitochondrial disease

Mitochondrial disease can be difficult to diagnose because it affects different organs and tissues in the body and patients have different symptoms. There is no single laboratory or diagnostic test that can confirm the diagnosis of mitochondrial disease. For this reason, referral to a medical facility that has a doctor specializing in these

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diseases is essential for diagnosis. Diagnosis begins with a series of tests and tests, including:

- Review of the patient's family history.
- Complete physical examination.
- Neurological examination.

Metabolic test: This includes blood and urine tests and, if necessary, a cerebrospinal fluid test (cerebrospinal fluid puncture).

Treatment of mitochondrial disease

There is no cure for mitochondrial disease, but treatment can help relieve symptoms and delay deterioration of health. Treatment depends on the patient and the specific mitochondrial disease diagnosed and its severity. However, there is no way to predict a patient's response to treatment or how the illness will affect the person in the long run. Two people do not respond to the same treatment in the same way, even if they have the same illness.