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Perspective

Protective Role of Mitochondrial Diseases and its Diagnosis

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DESCRIPTION

Mitochondrial disease is a long-term, often it is a hereditary disorder that occurs when mitochondria do not produce enough energy for the body to function properly. One in 5,000 people has hereditary mitochondrial diseases. Mitochondrial disease is a chronic (long-term) hereditary, often hereditary disorder that occurs when mitochondria do not produce enough energy to function properly. (Hereditary means that the disorder is inherited from the parent to the child.) Mitochondrial disorders can be present at birth, but can occur at any age. Mitochondrial disease can affect almost any part of the body, including cells in the brain, nerves, muscles, kidneys, heart, liver, eyes, ears, and pancreas.

Primary mitochondrial disease is a clinically heterogeneous group of diseases that results from dysfunction of the mitochondrial respiratory chain. The mitochondrial respiratory chain is the ultimate common pathway essential for aerobic metabolism. Tissues and organs that are heavily dependent on aerobic metabolism are preferentially involved in mitochondrial disease. Many hereditary and non-hereditary diseases are associated with mitochondrial mechanisms as a secondary feature. However, "primary mitochondrial disease" is defined as a known or suspected hereditary disease caused by a pathogenic variety of genes encoding the mitochondrial respiratory chain and related proteins.

CAUSES

Mitochondrial disease can be caused by mutations (acquisition or inheritance) in the mitochondrial DNA (mt DNA) or the nuclear genes that encode mitochondrial components. They can also be the result of acquired mitochondrial dysfunction due to drug side effects, infections, or other environmental causes. Oxalate can enter cells known to cause mitochondrial dysfunction.

Nuclear DNA has two copies per cell (excluding sperm and egg cells), one copy is inherited from the father and the other is inherited from the mother. However, mitochondrial DNA is inherited only from the mother (with some exceptions), and each mitochondria usually contains 2-10 mt DNA copies. During cell division, mitochondria randomly separate between two new cells. These mitochondria make more copies, usually reaching 500 mitochondria per cell.

Diagnosis of mitochondrial disease

Every 30 minutes, a child with mitochondrial disease is born by the age of 10. Overall, about 1 in 4,300 people in the United States have mitochondrial disease. Given the various potential symptoms that can occur, mitochondrial disease is difficult to diagnose and is often misdiagnosed. There are several ways to test if a person has mitochondrial disease. These include genetic diagnostic tests, genetic or biochemical tests on affected tissues such as muscle and liver, and other blood or urine-based biochemical markers.

TREATMENT

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.

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