Perry Syndrome

Subjects: Genetics & Heredity
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Perry syndrome is a progressive brain disease that is characterized by four major features: a pattern of movement abnormalities known as parkinsonism, psychiatric changes, weight loss, and abnormally slow breathing (hypoventilation). These signs and symptoms typically appear in a person's forties or fifties.

Keywords: genetic conditions

1. Introduction

Parkinsonism and psychiatric changes are usually the earliest features of Perry syndrome. Signs of parkinsonism include unusually slow movements (bradykinesia), stiffness, and tremors. These movement abnormalities are often accompanied by changes in personality and behavior. The most frequent psychiatric changes that occur in people with Perry syndrome include depression, a general loss of interest and enthusiasm (apathy), withdrawal from friends and family, and suicidal thoughts. Many affected individuals also experience significant, unexplained weight loss early in the disease.

Hypoventilation is a later feature of Perry syndrome. Abnormally slow breathing most often occurs at night, causing affected individuals to wake up frequently. As the disease worsens, hypoventilation can result in a life-threatening lack of oxygen and respiratory failure.

People with Perry syndrome typically survive for about 5 years after signs and symptoms first appear. Most affected individuals ultimately die of respiratory failure or pneumonia. Suicide is another cause of death in this condition.

2. Frequency

Perry syndrome is very rare; about 50 affected individuals have been reported worldwide.

3. Causes

Perry syndrome results from mutations in the *DCTN1* gene. This gene provides instructions for making a protein called dynactin-1, which is involved in the transport of materials within cells. To move materials, dynactin-1 interacts with other proteins and with a track-like system of small tubes called microtubules. These components work together like a conveyer belt to move materials within cells. This transport system appears to be particularly important for the normal function and survival of nerve cells (neurons) in the brain.

Mutations in the *DCTN1* gene alter the structure of dynactin-1, making it less able to attach (bind) to microtubules and transport materials within cells. This abnormality causes neurons to malfunction and ultimately die. A gradual loss of neurons in areas of the brain that regulate movement, emotion, and breathing underlies the signs and symptoms of Perry syndrome.

The Gene Associated with Perry Syndrome

- DCTN1

4. Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

In most cases, an affected person inherits the mutation from one affected parent. However, some cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

5. Other Names for This Condition

- parkinsonism with alveolar hypoventilation and mental depression

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