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Editorial

MYOTONIC DYSTROPHY: SYMPTOMS AND DIAGNOSIS

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EDITORIAL

Myotonic dystrophy is a muscular dystrophy that affects the muscles and other bodily systems and is hereditary. Muscle atrophy and weakness in the lower legs, hands, neck, and face are symptoms of myotonic dystrophy, which worsen over time. Myotonic dystrophy signs and symptoms commonly appear in people in their twenties or thirties. Myotonic dystrophy's severity varies greatly among persons who have it, even among family members.

The muscular loss and weakening that happens gradually advance to the point of impairment. Usually, it takes fifteen to twenty years for impairment to become severe once the symptoms start. When muscular weakness initially appears, it progresses more slowly and is less significant in those who are older.

Myotonic dystrophy is divided into two types:

- Type 1
- Type 2

Alterations (mutations) in two separate genes produce the two kinds. Type 2 myotonic dystrophy symptoms are frequently milder than Type 1 symptoms. At birth, a severe form of Type 1 myotonic dystrophy might be present. Congenital myotonic dystrophy is the name for this kind of Type 1 dystrophy. Only Type 1 myotonic dystrophy has been linked to congenital myotonic dystrophy, not Type 2.

Adult-onset myotonic dystrophy is the most frequent kind of muscular dystrophy. It affects around 1 in every 8,000 persons on the planet. In most countries, type 1 myotonic dystrophy is the most frequent kind. The prevalence of the two categories is determined by an individual's ethnic heritage. In persons of German heritage, Type 2 myotonic dystrophy is just as frequent as Type 1.

Symptoms

- Myotonic dystrophy causes gradual muscle wasting and weakening in people in their 20s and 30s. Muscle weakness and atrophy appear in their lower legs, hands, neck, and face. They also experience muscular stiffness and tightness (called myotonia), making it difficult for them to relax particular muscles after utilising them. One example of this difficulty is being unable to relinquish their grasp on a handshake or a doorknob.
- People with myotonic dystrophy suffer clouding of the lens in their eyes (cataracts) and anomalies in the electrical regulation of their pulse, in addition to muscular weakness and wasting (cardiac conduction defects).
- Myotonic dystrophy causes hormonal abnormalities in men, which can lead to baldness and the inability to father a child (infertility).
- Congenital myotonic dystrophy affects babies who are born with the signs and symptoms of myotonic dystrophy. They have flaws in every area of their lives.
- All of their muscles are weak, they have respiratory issues, and they suffer developmental delays, including mental impairment. These medical issues can sometimes be so severe that they result in death.

Diagnosis

A physical examination is used to diagnose myotonic dystrophy. A physical examination can detect muscular atrophy and weakness, as well as the existence of myotonia. A person with myotonic dystrophy may have a wasting aspect to their face, as well as jaw and neck muscular weakness. Males may experience frontal baldness.

To confirm the clinical diagnosis of myotonic dystrophy, a variety of laboratory tests can be employed. Electromyography (EMG) is a test that requires putting a tiny needle into the muscle. The electrical activity of the muscle is investigated, and it generally reveals distinct patterns of electrical discharge in the muscle.

A genetic test is the only way to know if you have myotonic dystrophy. A blood sample is obtained for this test in order to identify the changed gene (mutation) within the chromosomes of white blood cells. Myotonic dystrophy is caused by mutations in two genes: CNBP and DMPK. A mutation in the DMPK gene causes myotonic dystrophy Type 1. A mutation in the CNBP gene causes type 2 myotonic dystrophy. A small fragment of DNA is inappropriately repeated multiple times in each of these genes when it is mutated. The gene becomes unstable as a result of this aberrant repeat. These modifications prevent cells in muscles and other bodily tissues from working correctly, resulting in myotonic dystrophy signs and symptoms.

Treatment

Myotonic dystrophy presently has no cure or particular therapy. When muscular weakness worsens, ankle supports and leg braces might aid. There are drugs that can help with myotonia as well. Other symptoms of myotonic dystrophy can be addressed, such as heart difficulties and vision problems (cataracts).

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