Myotonic Dystrophy

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Myotonic dystrophy is part of a group of inherited disorders called muscular dystrophies. It is the most common form of muscular dystrophy that begins in adulthood.

genetic conditions

1. Introduction

Myotonic dystrophy is characterized by progressive muscle wasting and weakness. People with this disorder often have prolonged muscle contractions (myotonia) and are not able to relax certain muscles after use. For example, a person may have difficulty releasing their grip on a doorknob or handle. Also, affected people may have slurred speech or temporary locking of their jaw.

Other signs and symptoms of myotonic dystrophy include clouding of the lens of the eye (cataracts) and abnormalities of the electrical signals that control the heartbeat (cardiac conduction defects). Some affected individuals develop a condition called diabetes mellitus, in which blood sugar levels can become dangerously high. The features of myotonic dystrophy often develop during a person's twenties or thirties, although they can occur at any age. The severity of the condition varies widely among affected people, even among members of the same family.

There are two major types of myotonic dystrophy: type 1 and type 2. Their signs and symptoms overlap, although type 2 tends to be milder than type 1. The muscle weakness associated with type 1 particularly affects muscles farthest from the center of the body (distal muscles), such as those of the lower legs, hands, neck, and face. Muscle weakness in type 2 primarily involves muscles close to the center of the body (proximal muscles), such as the those of the neck, shoulders, elbows, and hips. The two types of myotonic dystrophy are caused by mutations in different genes.

There are two variations of myotonic dystrophy type 1: the mild and congenital types. Mild myotonic dystrophy is apparent in mid to late adulthood. Affected individuals typically have mild myotonia and cataracts. Congenital myotonic dystrophy is often apparent at birth. Characteristic features include weak muscle tone (hypotonia), an inward- and upward-turning foot (clubfoot), breathing problems, delayed development, and intellectual disability. Some of these health problems can be life-threatening.

2. Frequency

Myotonic dystrophy affects at least 1 in 8,000 people worldwide. The prevalence of the two types of myotonic dystrophy varies among different geographic and ethnic populations. In most populations, type 1 appears to be more common than type 2. However, recent studies suggest that type 2 may be as common as type 1 among people in Germany and Finland.

3. Causes

Myotonic dystrophy type 1 is caused by mutations in the *DMPK* gene, while type 2 results from mutations in the *CNBP* gene. The protein produced from the *DMPK* gene likely plays a role in communication within cells. It appears to be important for the correct functioning of cells in the heart, brain, and skeletal muscles (which are used for movement). The protein produced from the *CNBP* gene is found primarily in the heart and in skeletal muscles, where it helps regulate the function of other genes.

Similar changes in the structure of the *DMPK* and *CNBP* genes cause myotonic dystrophy type 1 and type 2. In each case, a segment of DNA is abnormally repeated many times, forming an unstable region in the gene. The gene with the abnormal segment produces an unusually long messenger RNA, which is a molecular blueprint of the gene that guides the production of proteins. The unusually long messenger RNA forms clumps inside the cell that interfere with the production of many other proteins. These changes prevent muscle cells and cells in other tissues from functioning normally, which leads to the signs and symptoms of myotonic dystrophy. If these changes affect the *DMPK* gene, the result is myotonic dystrophy type 1, if the *CNBP* gene is affected, the result is myotonic dystrophy type 2.

3.1. The Genes Associated with Myotonic Dystrophy

- CNBP
- DMPK

4. Inheritance

Both types of myotonic dystrophy are 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 has one parent with the condition.

As myotonic dystrophy is passed from one generation to the next, the disorder generally begins earlier in life and signs and symptoms become more severe. This phenomenon is called anticipation. The evidence for anticipation appears only in myotonic dystrophy type 1. In this form of the disorder, anticipation is caused by an increase in the length of the unstable region in the *DMPK* gene. A longer unstable region in the *CNBP* gene does not appear to influence the age of onset of myotonic dystrophy type 2.

5. Other Names for This Condition

- dystrophia myotonica
- myotonia atrophica
- myotonia dystrophica

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