

## The Myotonias

### What is myotonia?

Myotonia refers to the condition in which muscles are slow to relax after contracting. It occurs in a number of diseases such as myotonic dystrophy, myotonia congenita and paramyotonia congenita. However myotonic dystrophy is a very distinct condition in which myotonia is only a part. For this reason, and also because it is relatively common, there is a separate leaflet dealing specifically with this disorder. In the other two disorders myotonia is the main and often only symptom. They are the subjects of this leaflet.

### Myotonia congenita

The principal complaint in myotonia congenita is failure of muscle to relax normally after contracting; this can be severe enough to interfere with normal everyday activities. Myotonia most often occurs after long periods of rest, for example on waking in the morning, or on standing up and starting to walk after prolonged sitting. Most often it affects the limb muscles making walking and climbing stairs difficult. The face may also be affected and opening the eyelids can sometimes be difficult. Stiffness may not only occur after prolonged rest, but can often be brought on by cold, fatigue or emotional stress. It is relieved by exercise and is generally not accompanied by pain. One problem, which can be serious, is after a sudden movement, for example turning quickly to avoid traffic while crossing the road; the muscles tend to remain stiff and the individual may fall down as a result. Apart from myotonia, muscle enlargement, which may be pronounced, often occurs and affects the calves, thighs, shoulders and forearms.

### How is myotonia congenita inherited?

There are two types of myotonia congenita, both of which are inherited. The commoner type (the so-called Becker type) is inherited as an autosomal recessive trait (Becker myotonia congenita). In this case there is a 1 in 4 chance that any brother or sister may be affected, but affected individuals themselves are very unlikely to have affected children. In this form of myotonia, symptoms are often first noticed in late childhood and may then progress until adulthood when there may also be some degree of muscle weakness. Muscle enlargement can be marked.

The less common form is inherited as an autosomal dominant trait (Thomsen myotonia congenita) when there is a 1 in 2 chance of an affected parent transmitting the disease to any child he or she may have. In this form of myotonia, symptoms first appear in early childhood, often in infancy, and parents may notice that when their baby cries the eyes remain closed for an unusually long time. However, myotonia is mild and does not become more severe with time. Treatment is not usually necessary.

### What is the cause?

The underlying genetic fault in both the autosomal dominant and recessive forms of myotonia congenita causes a structural change in the channel (or pore) in the muscle cell wall that lets in chloride ions. Chloride ions help muscle to relax after contraction and the damaged chloride ion channels in myotonia congenita let in too few chloride ions for efficient relaxation.

### **Can it be treated?**

Many affected individuals learn to cope with their disability by working off the myotonia through repeated movements and avoiding the cold as much as possible. When severe, myotonia can be relieved by certain drugs (such as mexiletine) which can be prescribed by the doctor.

### **Paramyotonia Congenita**

In this condition myotonia evident from early childhood, is generalised but particularly affects the face and hands, and characteristically is aggravated by, or only occurs on exposure to, cold. It is not progressive and usually does not require treatment. Often muscle stiffness may actually be made worse by exercise, especially in cold weather. For this reason myotonia in paramyotonia congenita is referred to as "paradoxical" because repetitive activity often makes it worse rather than better. The condition is inherited as an autosomal dominant trait. Here the defect is in so-called sodium ion channels in the muscle membrane. A few patients may experience periodic paralysis—a sudden 'floppy' weakness of one or more limbs that can last minutes or hours. Episodes can be brought on by certain foods or drinks rich in potassium (e.g. bananas, orange juice), resting after exercise, missing meals or an infection.

### **Other forms of Myotonia**

Other rarer forms of myotonia have also been described, including one type in which prolonged attacks of weakness may occur. Because the myotonias are an uncommon group of disorders with differing severities and modes of inheritance, it is important to seek advice from a neurologist or medical geneticist with particular knowledge of these conditions.

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