

## Myotonic dystrophy

### What is myotonic dystrophy?

People with myotonic dystrophy, like those with other dystrophies, experience muscle weakness and wasting which is usually progressive. There are many differences, though, in the type of problem that people with myotonic dystrophy may have. These may include the following:

Types of muscles involved are usually in the face, jaw and neck area; the large, weight-bearing muscles of the legs and thighs are much less affected.

Rate of deterioration is commonly slow, with little change over a long period; some people never have significant muscle disability.

Muscle stiffness or 'myotonia' is characteristic, especially affecting the hands. Involvement of other body systems is frequent; associated problems may include cataracts, disturbance of heart rhythm, hormonal problems and, in children, learning difficulties.

Age at onset is very variable. Symptoms may appear at any time from birth to old age.

### How is myotonic dystrophy inherited?

This condition follows a 'dominant' inheritance pattern, which means that on average half of the children of an affected person are themselves affected. Both men and women are equally likely to be affected and to pass on the disorder, but affected women are more likely to have a severely affected child. In general (though not always) the disorder tends to be more severe in successive generations.

Most healthy adult relatives will not be likely to develop or pass on the disorder, but a careful assessment by an expert is important as mild features can easily be missed. Genetic testing on a blood sample for such relatives can now provide greater certainty, but should always be done with full information as part of genetic counselling. Genetic testing of healthy young children is not recommended.

Very few cases of myotonic dystrophy occur 'out of the blue'. Almost always, one parent proves to be affected, often very mildly. Some parents (or grandparents) prove to carry a very slight genetic change that will never give them symptoms. Careful study of the whole family often shows more members to be affected than would appear likely at first.

### What is the cause?

The changes in muscle and other body systems in myotonic dystrophy are now known to result from a specific genetic change (mutation) which in most cases involves a gene on chromosome number 19. The same change occurs in patients world-wide, but it is variable in extent, even in a single family, because it is unstable. The length of a particular 'triple repeat sequence' (CTG) is expanded in patients and this may vary from a slight expansion in mildly affected individuals to a very large one in severely affected children.

Until recently it has not been clear how genetic change causes the condition: the most likely mechanism is now thought to be that the expanded repeat is converted normally into the next stage (RNA), but then is unable to leave the cell nucleus. As a result of this trapping, a range of other types of RNA are affected, as are the protein they produce, which helps explain how a single genetic change can affect different body processes.

### **'PROMM' (proximal myotonic myopathy) and type 2 myotonic dystrophy.**

An important recent advance is the recognition of a second disorder with features resembling myotonic dystrophy. The muscle weakness tends to differ in distribution (more in proximal limb muscles, less in the face) and myotonia is often mild or absent. Cataract and heart involvement occurs as in myotonic dystrophy. It is now clear, that this condition (PROMM: proximal myotonic myopathy), is the same as some rare families thought, clinically, to have myotonic dystrophy but not showing the expected mutation and termed 'type2 myotonic dystrophy' or 'DM2'.

The gene involved has now been isolated on chromosome 3. Although quite different to that for myotonic dystrophy it contains a very similar, expand repeat (CCTG), which is likely to explain the clinical similarity of the two disorders. We are still learning about the details of this condition, but it is probably uncommon and accounts only for a small proportion of patients thought clinically to have myotonic dystrophy.

### **Future advances**

The research advances of the past 10 years have increased our understanding to the point where we can begin to see future possibilities for preventing or limiting the damage to muscle and other systems that occurs in myotonic dystrophy. In particular the genetic changes can now be re-produced in mouse models, which could allow the study of the effects of drugs and other agents that might be too untried to use safely on humans initially. It is difficult to predict how rapidly this work will progress, but possibilities exist now that were not present until very recently.

### **Problems and management**

Although no 'cure' for myotonic dystrophy exists at present, there is a lot that can be done to help those affected. Indeed, since many doctors are unfamiliar with the condition, it is essential that people who have myotonic dystrophy are themselves aware of the problems and dangers they may face. Some of these are mentioned here; of course they rarely all occur in one person, and many people have few symptoms, but it is important to be aware of them.

Operations and anaesthetics can be risky, even for mildly affected people. It is most important that any surgeon or anaesthetist should know a person has myotonic dystrophy before surgery is planned. Problems usually occur when doctors are unaware of the disorder; if care is taken, surgery is usually safe. A person may wish to wear a bracelet or locket stating their condition. A specific warning card is available that can be carried in a wallet. This can be obtained from the Myotonic Dystrophy Support Group (address below). 'Keep out of trouble' is a good motto for those with myotonic dystrophy.

A minority of people can develop heart problems, which are commonly treatable but can be serious if ignored. A regular cardiogram (ECG) is wise.

Some people who have myotonic dystrophy may have more trouble with other body systems than they do with their muscles. A symptom that appears quite unrelated may be connected. Excessive daytime sleepiness, swallowing difficulties and a range of bowel symptoms are examples. It is important that people with myotonic dystrophy should make sure that whoever treats them is aware that they have the condition and knows the wide range of associated problems.

If troublesome, muscle stiffness due to myotonia can be helped with certain drugs.

Children with myotonic dystrophy may have learning problems at a time when there are no muscle complaints. Again, be sure that myotonic dystrophy is borne in mind if this disorder is in the family. Affected women need careful management if undertaking a pregnancy. Not only is there a risk of a baby being severely affected, but problems in pregnancy and delivery may affect the mother.

Equipment for mobility and adaptations in the house can be very useful, though few affected people need a wheelchair. Weak neck muscles make a sound head-rest essential when driving.

### **In summary**

In summary, we now know a lot about myotonic dystrophy, but still have a long way to go. Helpful genetic counselling and family testing are now possible, but the best approach to treatment is to know about the condition, its risks and complications, and to be sure that your doctors do too.

### **Support Group**

Myotonic Dystrophy Support Group  
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