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# Myotonic dystrophy

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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 myotonic dystrophy patients 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 patients 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. Healthy family members are not likely to pass on the condition, but should be thoroughly checked by an expert since minor features can easily escape detection.

Very few cases of myotonic dystrophy occur 'out of the blue'. Almost always, one parent proves to be affected, often very mildly. Careful study of the whole family often shows more members to be affected than would appear likely at first.

## **What is the cause?**

The muscle of someone who has myotonic dystrophy shows characteristic changes under the microscope, and it is likely that a defect in the muscle cell membrane may

be responsible for both the muscle wasting and the myotonia (muscle stiffness). Myotonia on its own may occur in other genetic disorders, and is seen as a genetic defect in several animal species. Electrical studies show a characteristic disturbance leading to failure of muscle relaxation, and research again points to the muscle cell membrane as the site of the problem.

### **The myotonic dystrophy gene and mutation**

The gene responsible for myotonic dystrophy was identified at the beginning of 1992, largely by British research workers funded by the Muscular Dystrophy Campaign. The defect has proved to be a region of unstable genetic material (DNA) on chromosome 19. This unstable sequence can show an expansion that varies from slight to very large in different people, and which can now be used as a specific test for the disorder, both for detecting gene carriers and in prenatal diagnosis.

The new direct test for the gene is likely to be more accurate than previous tests, and can be undertaken in those with no clear family history or living affected relative. However, its use should be accompanied by full genetic counselling and clinical assessment to avoid a misleading situation.

### **Future advances**

The isolation of the myotonic dystrophy gene is likely to lead to rapid progress in research. The gene itself normally produces a previously unknown 'protein kinase' that is likely to be important in membrane function of muscle and other organs. The discovery should open the way to a greater understanding of the disease overall, and in the longer term may give possibilities for treatment.

### **Problems and management**

Although no 'cure' for myotonic dystrophy exists at present, there is a lot that can be done to help those affected. Since many doctors are unfamiliar with the condition, it is essential that people who have myotonic dystrophy are themselves aware of the problems and risks they may face. Some of these are mentioned here; 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 patient 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. Patients 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. A regular cardiogram (ECG) is wise.

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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. It is important that people with myotonic dystrophy should make sure that whoever treats them is aware that they have the conditions 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**

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.

The UK based Myotonic Dystrophy Support Group is affiliated to the Muscular Dystrophy Campaign. It offers support and encouragement to families and individuals affected by myotonic dystrophy. You can contact them at:

### **Myotonic Dystrophy Support Group**

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