Myotonic Dystrophy
SUPPORT GROUP

Myotonic Dystrophy and the Brain

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Myotonic Dystrophy

DM1 is a multi-system disorder and not just a muscle disorder. The additional features that can develop have been recognised for many years and may include diabetes, cataracts, swallowing difficulty, changes in heart rhythm, sleepiness and bowel problems.

It has long been known that children severely affected at birth often have learning difficulties. In adults, changes in personality as well as sleepiness have also been recognised for many years but have only more recently been the subject of intensive research. This article describes these changes and current areas of research.

Families of affected adults have always been clear that there are changes in personality. The exact nature of the changes has been very hard to describe. In the past imprecise terms such as 'apathetic' or 'lethargic' were used and 'socialisation difficulties' were described.

MD

There are many reasons why someone with myotonic dystrophy may show changes in personality. These complicating factors include:

- any drugs with sedating effects can cause a change in behaviour
- muscle weakness can cause a change in facial appearance, which people may perceive as looking emotionless or even 'dopey'

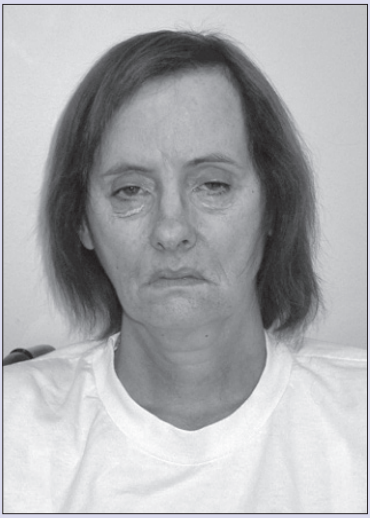

Figure 1: *Photograph of a lady with myotonic dystrophy, showing weakness of the facial muscles.*

(Reproduced from Turner, Hilton-Jones. J Neurol Neurosurg Psychiatry 2010).

- it is known that sleep patterns change in DM1 and daytime sleepiness will cause people to be drowsy and have less energy. They may then find it hard to concentrate
- having a long term progressive illness is very hard to cope with and many people with such a diagnosis have periods of low mood or depression, leading to withdrawal from activity
- changes in the voice which may occur in DM1 can also affect how people are understood by others.

It has been hard to show that there are additional effects on the brain in DM1 which are due to the disease. We now believe this to be the case and our understanding of the difficulties is improving.

Psychological research has shown there may be changes in personality. In addition, changes have also been described on brain scans and microscope studies.

- studies which have tried to make allowance for the complicating issues described above have shown

  - problems with 'executive function' - difficulties in planning and sorting complex tasks

  - problems maintaining concentration

  - avoidant traits

  - difficulties in interpreting subtle social "clues" such as facial expression to infer what other people are thinking or feeling

- MRI scans have shown changes

  - abnormalities in the 'wires' of the brain that connect areas with differing functions

  - reduction in the size of the brain over time (more than that associated with normal ageing)

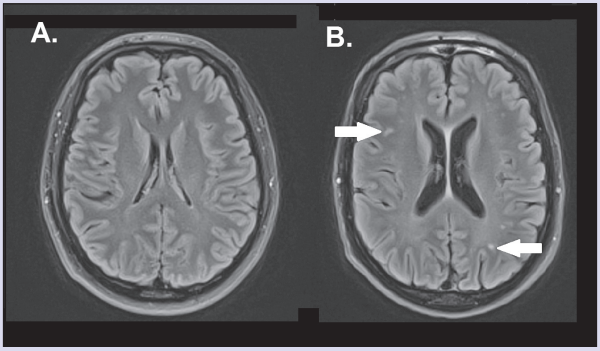

Figure 2: *MRI of brain in two people of similar ages, one with a diagnosis of myotonic dystrophy (B) and one without (A). Scan B shows some widening of the dark, fluid-filled spaces in the centre of the brain, indicating loss of brain volume. In addition, changes affecting the white matter (marked with arrows) are seen more frequently in the affected person.*

- pathology studies on brain have shown changes including accumulation of structural proteins / building blocks within cells.

We now believe these lines of evidence indicate that the brain is involved in DM1. It remains unclear precisely how these changes relate to one another, and the extent to which an individual with myotonic dystrophy may experience these difficulties varies considerably.

If severe, they can have impacts on relationships, employment and ability to comply with medical care - over and above those due to the muscle weakness. The reasons for variability in these symptoms is not well understood although they do, to some extent, relate to the severity of any muscle weakness and the expansion size within the myotonic dystrophy gene. They do evolve over time. There remains disagreement over the relationship between the severity of the MRI changes and the psychological problems.

We are not yet in a position to cure the basic mechanisms by which DM1 affects the brain or other organs, but there are approaches which may help some people

- modafinil is a drug sometimes used to treat daytime sleepiness. It may improve alertness but has significant side-effects and so may not be appropriate for all
- It is important to treat any associated sleep disorders at night as well as any heart rhythm problems. There is evidence that both of these factors may contribute to the psychological difficulties
- it is important to recognise and treat depression

In addition, trying to maintain an active lifestyle including gentle exercise as symptoms allow, and participation in social activities is very important. Muscle symptoms and fatigue can make it tempting to avoid such activities, but a sedentary lifestyle and social isolation can in fact

have a negative impact on energy levels, mood and motivation. The use of a psychology-based technique called Cognitive Behavioural Therapy (CBT), aiming to increase activity levels and social participation among people with DM1, is currently the subject of a clinical trial (the OPTIMISTIC Study; optimistic-dm.eu), which is ongoing at the time of writing.

Other current areas of research interest include looking at how changes seen on brain scans change over time, and in turn how changes seen in specific regions of the brain relate to symptoms such as excessive sleepiness or difficulty with certain aspects of thinking. Crucially, researchers are also working to identify the most meaningful ways to measure "brain-related" symptoms, so that any effect of potential treatments on these symptoms can be accurately assessed in a drug trial.

Further information about Myotonic Dystrophy can be obtained from:

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Email: contact@mdsguk.org
www.**myotonic**dystrophy**support**group.org

Other publications available from the Myotonic Dystrophy Support Group:

- Anaesthesia and Sedation for patients with Myotonic Dystrophy
- Basic Information for Midwives
- Bowel Problems in Myotonic Dystrophy
- Congenital Myotonic Dystrophy
- Excessive Daytime Sleepiness and Myotonic Dystrophy
- Facts for patients, family members and professionals
- Myotonic Dystrophy and the Eye
- Myotonic Dystrophy Support Group
- Relatives Information
- The Heart and Myotonic Dystrophy
- Why do we get new families with Myotonic Dystrophy?

Myotonic Dystrophy
SUPPORT GROUP

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