DM1 is the most common adult muscular dystrophy worldwide. It is a complex and variable disease that effects people in many different ways. The condition affects multiple organs and multiple systems within the body.

Symptoms include:

- Muscle weakness (e.g. in the face, neck, hands and forearms);
- Myotonia (cramping, difficulties in releasing grip, etc.);
- Trouble swallowing;
- Cataracts (blurry or cloudy vision) and other eye problems;
- Heart Conditions (e.g. arrhythmia);
- Tiredness and daytime sleepiness;
- Problems with planning and thinking.

Why DM1?

The internationally agreed abbreviation for myotonic dystrophy type 1 is DM1. This abbreviation comes from the Latin name *dystrophia myotonica* and is used to prevent confusion with MD a term used for all muscular dystrophies.

Management of DM1

DM1 affects thousands of people across Europe and globally, however there is currently no treatment to slow down, prevent or cure most of the symptoms. Appropriate care and support can make a great difference to help those affected. It is recommended that adults with the condition should see a neurologist at least once a year and in addition appropriate treatment should be provided for different symptoms; such as seeing a cardiologist for heart conditions or an ophthalmologist for eye problems.

A meeting of international experts was held in 2009 which highlighted the need for improved standards or care and treatment for people with DM1. OPTIMISTIC hopes to further highlight the need for better care and suggest innovative ways to treat and improve the management of patients with DM1. You can download the report here.

Genetics of DM1

DM1 is an autosomal dominant inherited disease. This means that if you carry a specific mutation there is a 50 % likelihood it will be passed on to your children. DM1 is caused by the repetition of a certain segment of the DMPK gene. This repeat causes the expansion of the

DNA making it unstable; the triplet repeats is found about 35 times in an unaffected person but expands from over 50 to several thousand in people with DM1. There is generally a correlation between the number of repeats and the severity of the disease. However patients with the same number of repeats can be very differently affected.

DM1 also follows a phenomenon known as “anticipation” in which the repeat number increases with generation. For example a grandparent with DM1 will have a lower repeat number than their grandchildren. This means that younger generations are often more severely affected and earlier in life.

For more information about organisations that provide help and support for people with myotonic dystrophy please go to the “help and support” section of the website.

OPTIMISTIC is focusing on myotonic dystrophy type 1 (DM1), however it also exists in type 2 (DM2). You can find out more about myotonic dystrophy type 2 here.