optimistic

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Welcome to the second OPTIMISTIC newsletter. You are receiving this newsletter as we think you might find it interesting. To continue to receive updates about this project then please sign up for the newsletter at www.optimistic-dm.eu or contact Libby Wood elizabeth.wood2@ncl.ac.uk.

There are many different aspects to OPTIMISTIC and throughout the course of the project we will try and introduce you to them all. In this issue we focus on the genetic analysis that is being carried out as part of the project, as well as providing an update on the progress of the study.

Thank you for your interest in OPTIMISTIC!

What is OPTIMISTIC?

OPTIMISTIC is a 4 year EU-funded research project focusing on myotonic dystrophy type 1 (DM1). The project will form a trial testing a new non-pharmacological treatment for the condition. A combination of cognitive behavioural therapy and exercise will be assessed to see if it can increase activity, reduce fatigue, and improve quality of life.

The trial is being carried out in four cities across Europe, Newcastle; United Kingdom, Nijmegen; Netherlands, Munich; Germany and Paris; France.

As well as the trial itself we are trying to find how and why myotonic dystrophy affects people in different ways. Researchers across Europe are looking at a number of different aspects of the condition including biomarkers, genetics and outcome measures.

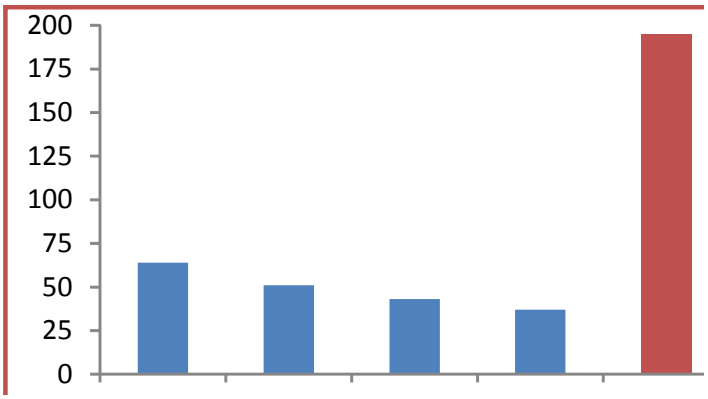
We will give you more information about these different areas through these newsletters. You can find past newsletters and additional information on the project website: www.OPTIMISTIC-dm.eu

Baziel Van Engelen is coordinating the project from Nijmegen in the Netherlands.

Progress Update

Recruitment

OPTIMISTIC started recruiting participants in April 2014; the 100th participant was randomised in October 2014. As of Feburary 2015 we have 190 people included in the study.



On average over 17 people have joined the study each month, 64 in Munich, 51 in Nijmegen, 43 in Newcastle and 37 in Paris. By April 2015 we hope to have more than 200 participants involved across the four sites.

Substudy Leg MRI

The first particpans have been included in to sub study looking at the muscles in the leg. This study will only take place in Nijmegen and Paris. uUp to 50 participants will be included in this substudy and will have scans of their leg taken at the beginning and end of the OPTIMISTIC study. More information about the leg MRI is available on the website;
www.optimisitc-dm.eu

Substudy Cardiac MRI

The first participants have now been included into a sub study looking at the heart. Forty patients in total will be included in this study; they will have an MRI scan of their heart at the beginning and end of the OPTIMISTIC study. More information about this cardiac study is available on the website;
www.optimisitc-dm.eu

Biobank

Blood and urine samples are taken from all participants taking part in the study. All of these samples are being sent to the Newcastle Biobank for Research of Neuromuscular Disorders. This means that other researchers, if they have received ethical approvals, will be able to use these samples to find out more about myotonic dystrophy. No identifiable (e.g. your name or contact details) information are given to researchers working on your samples.

Diary Dates

28th February 2015
Rare Disease Day, Worldwide
6th-7th June 2015
OPTIMISTIC steering committee meeting, Paris, France
8th- 12th June 2015
IDMC-10, international myotonic dystrophy congress, Paris, France.
30th September- 4th October 2015
World Muscle Society, Brighton, UK

Thank you for reading our Newsletter. Any comments or feedback you have about this newsletter would be most appreciated – please let us know what is useful, what is not and what you feel is missing so that we can improve the next edition. Contact Libby Wood; Elizabeth.wood2@ncl.ac.uk

Spotlight on Genetics.

As part of the OPTIMISTIC study blood is taken from participants at three time points. This blood will be used for both genetic and biomarker studies. Here we will tell you more about the genetic studies.

For genetic studies DNA is extracted from the blood. DNA is often compared to a recipe, as it contains the instructions needed to construct other components of cells, such as proteins which allow our bodies to function. DNA is made up of just four molecules known as bases; these bases are represented by the letters A, C, T and G. It is these bases that form genes; sections of DNA that contain instructions on how to make a specific protein.

We know that the symptoms of myotonic dystrophy type 1 are caused by a change in DNA (a mutation) that occurs in a gene called DMPK. This change is an expansion made up of a series of CTG bases repeated multiple, potentially hundreds or thousands, of times. This triplet repeat expansion is not the same size in everyone with DM1 and it is not always the same in all cells in the body. The expansion can also change throughout a person's lifetime, so it is not the same when you are? 40 years old as when you were born. The variability of this expansion makes it difficult for doctors to know how myotonic dystrophy will progress in an individual.

The DNA that is extracted will be analysed by Professor Darren Monckton and his team at Glasgow University. This team will try and see if there is a relationship between the number of CTG repeats and the age of onset of the disease as well as if there is a link to the severity of the disease. This will increase the understanding of the genetic mechanism in myotonic dystrophy and how it relates to the symptoms people experience. It is hoped that the samples collected in OPTIMSITIC will help Prof Monckton work towards a prognostic tool that can help estimate how the CTG repeat will change and throughout a person's lifetime and how this will affect symptoms.

Some cases of myotonic dystrophy have shown special sequences of bases in the CTG repeat expansion. We will look to see if any of these repeats are present in the samples collected in OPTISMITC and if this affects the symptoms that are presented.

If you are interested in hearing more about the genetics of myotonic dystrophy professor Prof Monckton recorded a webinar for the Myotonic Dystrophy Foundation which is available to listen to on their website. www.myotonic.org/webinar-everything-you-wanted-know-about-ctg-repeats.