A RETROSPECTIVE STUDY OF SUBJECTS WITH MUTATIONS IN THE C12ORF65 GENE CAUSING COMPLEX CHARCOT-MARIE-TOOTH DISEASE TYPE 6 (CMT6): LAY SUMMARY

WHAT IS THIS STUDY ABOUT?

The aim of this study is to better understand the clinical appearance and course of Charcot-Marie-Tooth disease type 6 (CMT type 6) caused by a genetic change ('mutation') in the C12orf65 gene. The first symptom in individuals with C12orf65 deficiency is usually childhood-onset optic atrophy, which is followed by weakness in the arms and legs (caused by neuropathy), ataxia and in some people, learning difficulties.

In the era of emerging clinical trials in rare genetic diseases it is important to capture the full clinical picture, as we need to develop outcome measures to detect the effect of any trial drugs. The purpose of this study is to better understand the natural history and progression of disease caused by mutations in C12orf65, by building a database of clinical data on these participants. This will help us to guide the design of innovative new clinical trials, with the long-term goal of developing treatments for patients.

WHO IS RUNNING THIS STUDY?

The Chief Investigator is a doctor called Professor Rita Horvath, who carries out specialist research into neurodegenerative diseases at the University of Cambridge and Addenbrooke's Hospital.

WHAT WILL IT INVOLVE?

As a participant in this study, you would be asked to take part in a virtual study visit by telephone or online, to collect clinical data. We are particularly interested in collecting information related to your (or your family members') genetic diagnosis, medical history, family history, birth and development history, and examination, imaging or laboratory results. It is expected that this study visit will take approximately 1-1.5 hours. If you have any copies of medical notes, clinic letters or test results, we will also ask if you would be willing to share these with the study team.

WHO ARE WE RECRUITING?

We are recruiting individuals of any age with a confirmed (or likely) C12orf65 mutation.

WANT TO LEARN MORE?

If you think you or a family member might be eligible and are interested in taking part in this research, or would like any further information, please contact the study team directly.

Email: add-tr.mitoteam@nhs.net
Tel: 01223 331506