Lay Summary

BRAIN UK Ref: 18/007

Charcot-Marie-Tooth Disease and related disorders: A Natural History Study

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CharcotMarieTooth Disease (CMT) is the most common cause of inherited peripheral neuropathies affecting approximately 1 in 2500 people. People with this condition present with upper and lower limb weakness, wasting and sensory loss as a result of degeneration of the long peripheral nerves supplying the muscles. Despite the clinical similarities among patients with CMT the group is genetically very different.

Advances have been made in identifying the genes that cause CMT. However, the best way to treat the different variants of this disorder is not known. In addition, we don't have natural history data of most forms of inherited neuropathies.

We are currently doing a wider study to characterise the features of different types of CMT, the long term progression of the disease and how some specific gene mutations can cause the neuropathy. We will supplement the study with some cases of CMT from BRAIN UK.