**Research Project:**

# A feasibility study of Remote Patient-Reported Outcome measures in Mitochondrial disease and Spinocerebellar ataxias (R-PROMS)

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## Scientific summary

There are many causes of cerebellar ataxia, including rare genetic diseases such as primary mitochondrial disease (PMD), spinocerebellar ataxias (SCAs) and autosomal recessive cerebellar ataxias (ARCAs). Symptoms of ataxia including walking difficulties due to lower limb incoordination causing impaired balance, upper limb ataxia causing impaired hand and finger dexterity, slurred speech, swallowing difficulties and abnormal eye movements caused by progressive cerebellar degeneration.

Patients with rare diseases often encounter obstacles to participating in clinical research due to geographic barriers of accessing specialist and research team, and often time-demands study protocols for data collection during site visits. Moreover, many observational and interventional studies of PMD and SCAs have focused on ambulatory patients and excluded patients who have more advanced diseases. Since the COVID-19 pandemic, the decentralised trial design using various methods of electronic assessment is attracting more attention in multiple medical fields. This study will involve the recruitment of participants with either a diagnosis of mitochondrial disease, a diagnosis of any subtype of spinocerebellar ataxia or autosomal recessive cerebellar ataxia, who will be asked to attend the study site visit in Newcastle upon Tyne for a baseline assessment, following by the completion of study assessments online, including cognitive motor tasks, speech assessment, cognitive tasks and a range of patient reported outcome measures (PROMS) every two months for the period of one year.

This study aims to widen research participation for people with ataxia in the UK irrespective of their disease severity and genetic causes and aims to provide knowledge that can be translated into clinical practice and clinical trial design.

## Lay summary

Ataxia refers to symptoms including unsteadiness and walking difficulties, incoordination of the arms and legs, and slurred speech. Ataxia is caused by degeneration of part of the brain called the cerebellum, and there are many causes including genetic diseases such as spinocerebellar ataxia, primary mitochondrial disease, and autosomal recessive cerebellar ataxias. There are significant barriers for patients with ataxia involved in taking part in clinical research, including travelling distances, financial and caregiver burden, and restrictive inclusion criteria specified by

individual research projects that include participants with milder disease burden. Since the COVID-19 pandemic, it is becoming apparent that the application of digital technologies in clinical practice and research helps remove physical barriers from patients who require repeated assessments.

This project aims to study symptoms of ataxia and the quality of life of adult patients with a diagnosis of mitochondrial disease, a diagnosis of any type of spinocerebellar ataxia or autosomal recessive cerebellar over one year. A face-to-face visit is planned for a physical examination at the beginning of the study. The participants will complete several online assessments focusing on finger movements, reaction time, speech and questionnaires related to ataxia and quality of life. The online assessments will be repeated every two months. This is a research collaboration between Newcastle University, UK, and Monash University, Australia. Funding from Ataxia UK offers the research opportunity to patients who do not usually take part in research studies due to severe physical disability, as well as those with very rare genetic causes.

At the end of the study, the research findings will provide evidence that online repeated assessments are suitable and reliable for identifying how ataxic symptoms change over time, which will help inform the design of clinical trials.