Lay Summary

BRAIN UK Ref: 18/011

Investigation of cerebellar ataxia

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Cerebellar ataxia is a neurological problem in which patients complain of poor balance, have movement problems, demonstrate abnormal eye movements and also have speech and swallowing problems.

For a proportion of these ataxias there is a known fault in the genetic make-up which is the reason why this neurological problem develops in these patients, but for some the underlying cause is not known.

We have recently identified a new genetic defect in a small group of patients with ataxia. We want to investigate if muscle and nerve biopsies from these patients can tell us anything important about the underlying mechanisms. We want to see if there are any correlations with the pathological alterations observed in these patients and those without the genetic defect.

We hope that this study will help us to understand further the underlying mechanisms leading to ataxia and neuropathy development, which we hope will help diagnose the disease more accurately.

Publications:

Date	Publication title
2019	Biallelic expansion of an intronic repeat in RFC1 is a common cause of late-onset ataxia.