

Disruption of balance in spinocerebellar ataxia: Developing targeted therapies

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Scientific abstract

Disruption of balance is a common and devastating symptom of spinocerebellar ataxia (SCA). It can result in falls and reduced quality of life. Our purpose here is to enable the development of novel therapies that will improve balance control of people with SCA. However, the widespread and variable areas of pathology in SCA lead to multiple impairments that have the potential to disrupt balance in many ways. In order to develop targeted therapies we intend first to develop diagnostic tests that can identify the disrupted balance subprocesses involved with sensory processing, multisensory integration, and motor control. We shall initially apply these tests to selected patients with SCA6 who present with relatively pure cerebellar pathology. The procedure will be repeated on patients with SCA1-3 who present with cerebellar and extra-cerebellar pathology. We shall correlate the laboratory data and the subjects' phenotype, using a new validated rating scale for cerebellar ataxia (SARA), with their self-reported fall history to identify the dominant impairment associated with imbalance. With these data we will design a therapeutic intervention, which we will pilot on a small selected group of patients with SCA6. It is expected that this will generate sufficient information to power a larger future study.

Lay summary

Falls occur in up to 93% of people with spinocerebellar ataxia (SCA). It often results in injury leading to a fear of falling, a restriction of everyday activities and a reduced quality of life. The falls are mainly caused by balance difficulties. The long-term aim of this project is to start developing novel therapies that will improve the balance control of people with different types of SCA. However, before we can develop therapies we need to understand the underlying causes of the individual's balance problem. The complexity of balance control coupled with the variability of the symptoms in different types of SCA means there are many possible reasons for balance problems. Therefore our shorter-term aims are to develop a series of laboratory tests, clinical tests and questionnaires that will help us to understand precisely how balance is disrupted in different types of SCA. We will then use this information to design one physiotherapy intervention targeted at the most common impairment, and we will pilot its effectiveness in a small selected group of patients with SCA6.

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