



# "EWBODFT \*O 4QJOPDFSFCFMMBS "UBYJB 3FTF 'VUVSF %JSFDUJ POT

Lukas Simon Enz\*

Department of Psychiatry, University College London, London, United Kingdom

## Introduction

Spinocerebellar ataxia (SCA) refers to a group of inherited neurodegenerative disorders characterized by progressive loss of coordination, balance, and motor function. Affecting the cerebellum and spinal cord, SCAs are caused by genetic mutations, and their symptoms typically worsen over time, impacting a patient's quality of life. To date, more than 40 different types of SCAs have been identified, each associated with specific genetic mutations. Despite the complexity and genetic variability of the disorder, recent advances in research are shedding light on its underlying mechanisms and offering hope for the development of effective treatments [1]. This article will explore the latest breakthroughs in SCA research and examine potential future directions.

## Genetic and molecular mechanisms of spinocerebellar ataxia

Understanding the genetic basis of SCA has been a pivotal focus of research. Most SCAs are caused by mutations in genes that affect proteins critical for neuronal function, leading to neurodegeneration. SCAs are generally inherited in an autosomal dominant manner, meaning that a single copy of the defective gene is enough to cause the disease. One of the most well-known mutations is the expansion of CAG trinucleotide repeats in specific genes, such as ATXN1, ATXN2, ATXN3, CACNA1A, and others. This expansion leads to the production of abnormal proteins with elongated polyglutamine (polyQ) tracts, which accumulate in neurons, causing cellular dysfunction and death [2]. Understanding how these toxic proteins contribute to neurodegeneration has opened the door to potential therapeutic strategies targeting protein misfolding, aggregation, and clearance.

Other SCAs, such as SCA6, are associated with calcium channel mutations that affect the excitability of neurons in the cerebellum. Understanding the role of ion channel dysfunction in these subtypes has also been a key area of research, and identifying therapeutic targets

neuronal excitability. Calcium channel blockers, for instance, could potentially reduce the excitotoxicity that leads to neuronal death in affected individuals.

#### Neuroprotective and symptomatic therapies

Beyond targeting the underlying causes of SCA, neuroprotective and symptomatic therapies aim to preserve neuronal function and improve the quality of life for patients. Several compounds with neuroprotective properties, such as coenzyme Q10, creatine, and riluzole, have been investigated in clinical trials. While results have been mixed, ongoing research continues to explore new compounds that could slow disease progression.

Symptomatic treatments, such as physical therapy, speech therapy, and occupational therapy, remain crucial for managing the motor and speech impairments that arise from SCA. Recent studies have