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Introduction

Spinocerebellar ataxia (SCA) refers to a group of inherited neurodegenerative disorders characterized by progressive loss of coordination, balance, and motor function. A ecting 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 di erent types of SCAs have been identi ed, each associated with speci c genetic mutations. Despite the complexity and genetic variability of the disorder, recent advances in research are shedding light on its underlying mechanisms and o ering hope for the development of e ective treatments [1]. is 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 a ect 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 speci c genes, such as ATXN1, ATXN2, ATXN3, CACNA1A, and others. is 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 a ect the excitability of neurons in the cerebellum. Understanding the role of ion channel dysfunction in these subtypes has also beeUndhhcellup 0.de tiafyng the apeutic sargetis