CANVAS

cerebellar ataxia, neuropathy, vestibular ataxia syndrome (CANVAS) literature, providing both clinical and genetic insights that might facilitate the timely clinical and genetic diagnosis of this disease.

Recent findings: Recent advancements in the range of the clinical features of CANVAS have aided the development of a broader, more well-defined clinical diagnostic criteria. Additionally, the identification of a biallelic repeat expansion in RFC1 as the cause of CANVAS and a common cause of late-onset ataxia has opened the door to the potential discovery of a pathogenic mechanism, which in turn, may lead to therapeutic advancements and improved patient care.

Summary: The developments in the clinical and genetic understanding of CANVAS will aid the correct and timely diagnosis of CANVAS, which continues to prove challenging within the clinic. The insights detailed within this review will raise the awareness of the phenotypic spectrum and currently known genetics. We also speculate on the future directions of research into CANVAS [1)]

1)
Sullivan R, Kaiyrzhanov R, Houlden H. Cerebellar ataxia, neuropathy, vestibular areflexia syndrome: genetic and clinical insights. Curr Opin Neurol. 2021 Aug 1;34(4):556-564. doi: 10.1097/WCO.0000000000000961. PMID: 34227574.