Special Issue

Cerebellar Ataxia with Neuropathy and Vestibular Areflexia Syndrome (CANVAS): A Multisymptomatic Neurologic Disease

Message from the Guest Editor

Initial descriptions of CANVAS (cerebellar ataxia, sensory neuronopathy and vestibular areflexia) were reported in the 1990s, its genetic defect was not found until 2019, when two groups described the expansion of the pentanucleotide AAGGG in intron two of the RFC1 gene responsible for most cases of CANVAS. Others pathogenic and non-pathogenic expansion, as point mutations, has been recently described in the RFC1gene. The underlying pathogenic mechanisms of RFC1 expansions have not been resolved yet. CANVAS is a multisystem syndrome, wherein sensory neuropathy is one of the most consistent findings, accompanied with cerebellar and vestibular dysfunction symptoms. Patients harboring a biallelic expansion in the RFC1 gene may also present with small fiber neuropathy, autonomic symptoms, chronic cough, cognitive impairment, and Parkinsonism. This Special Issue aims to provide readers with comprehensive and up-to-date studies pertaining to basic and clinical research in CANVAS that unravel the pathogenic mechanisms and clarify the clinical manifestations in CANVAS, potentially enhancing the patients' quality of life.

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Deadline for manuscript submissions

closed (30 April 2024)

Brain Sciences

an Open Access Journal by MDPI

Impact Factor 2.8
CiteScore 5.6
Indexed in PubMed

mdpi.com/si/184491

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