DISEASE:
Adult-onset autosomal recessive cerebellar ataxia

NAME:	Adult-onset autosomal recessive cerebellar ataxia
DESCRIPTION:	A rare, genetic, autosomal recessive cerebellar ataxia disease characterized by adulthood-onset of slowly progressive spinocerebellar ataxia, manifesting with gait and appendicular ataxia, dysarthria, ocular movement anomalies (e.g. horizontal, vertical, and/or downbeat nystagmus, hypermetric saccades), increased deep tendon reflexes and progressive cognitive decline. Additional variable features may include proximal leg muscle wasting and fasciculations, pes cavus, inspiratory stridor, epilepsy, retinal degeneration and cataracts. Brain imaging reveals marked cerebellar atrophy and electromyography shows evidence of lower motor neuron involvement.
ORPHACODE:	284289
SYNONYMS:	Autosomal recessive spinocerebellar ataxia type 10 SCAR10
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ANO10
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/2018

RELATED CONTENT

Related Analytes

- anoctamin 10

Source URL: http://gentest.healthdata.be/disease/2018