Opis choroby *

Definicja

Spinocerebellar ataxia type 3 (SCA3), also known as Machado-Joseph disease, is the most common subtype of type 1 autosomal dominant cerebellar ataxia (ADCA type 1; see this term), a neurodegenerative disorder, and is characterized by ataxia, external progressive ophthalmoplegia, and other neurological manifestations.

Dane

Klasyfikacja
Choroba

Synonimy
Azorean disease of the nervous system
Autosomalnie dominujące zwyrodnienie nigrostriatalne
Azorańska choroba układu nerwowego
Choroba Machado
Choroba Machado i Josepha
MJD
MJD
Machado disease
Machado-Joseph disease
Nigro-spino-dentatal degeneration with nuclear ophthalmoplegia
SCA3

Kod ORPHA
98757

Kod OMIM
109150

Kod ICD10
G11.8

Kod ICD11
8A03.16

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*Źródło

orphanet