Opis choroby *

Definicja

A rare, genetic, paroxysmal dystonia disorder characterized by childhood to adolescent-onset of episodic paroxysmal choreoathetosis, triggered mainly by sudden movements, prolonged exercise, anxiety and emotional stress, in association with progressive spastic paraparesis (onest in adulthood), gait ataxia, mild to moderate cognitive impairment, and/or epileptic seizures. Episodes typically last from a few minutes to hours, have a variable frequency (daily to yearly), and are relieved by rest. Frequency of episodes tends to decrease with age.

Dane

Klasyfikacja
Choroba

Synonimy
DYT9
Choreoatetoza/spastyczność epizodyczna
DYT9
Episodic choreoathetosis/spasticity

Kod ORPHA
53583

Kod OMIM
601042

Kod ICD10
G24.8

Kod ICD11
8A02.2

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*Źródło

orphanet