Opis choroby *

Definicja

A rare genetic movement disorder characterized by dystonia affecting at first an upper limb, less frequently beginning in the head and neck region, before slowly spreading to other locations. The clinical spectrum, like age of onset, is variable with focal, segmental, or generalized distribution, but cranial involvement with speech difficulties and cervical involvement are typical, whereas lower limbs are often spared. With progression of the disease, many patients suffer from generalized dystonia while mostly remaining ambulatory.

Dane

Klasyfikacja
Choroba

Synonimy
DYT6
Dystonia uogólniona rozpoczynająca się od szyi i kończyn górnych
DYT6
Idiopatyczna dystonia torsyjna typu mieszanego
Generalized cervical and upper-limb-onset dystonia
Idiopathic torsion dystonia of mixed type

Kod ORPHA
98806

Kod OMIM
602629

Kod ICD10
G24.1

Kod ICD11
8A02.0Y

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*Źródło

orphanet