

Opis choroby *

Definicja

Dopa-responsive dystonia (DRD) describes a group of neurometabolic disorders characterized by dystonia that typically shows diurnal fluctuations, that responds excellently to levodopa (L-dopa) and that is comprised of autosomal dominant dopa-responsive dystonia (DYT5a), autosomal recessive dopa-responsive dystonia (DYT5b) and dopa responsive dystonia due to sepiapterin reductase (SR) deficiency.

Dane

Klasyfikacja

Grupa fenomenów

Synonimy

HPD with diurnal fluctuation

Dziedziczna postępująca dystonia z wahaniami dobowymi

HPD z wahaniami dobowymi

Hereditary progressive dystonia with diurnal fluctuation

Kod ORPHA

255

Kod OMIM

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Kod ICD10

G24.8

Kod ICD11

8A02.11

*Źródło

orphanet