

Opis choroby *

Definicja

A muscular channelopathy presenting with a pure myotonia dramatically aggravated by potassium ingestion, with variable cold sensitivity and no episodic weakness. This group includes three forms: myotonia fluctuans, myotonia permanens, and acetazolamide-responsive myotonia.

Dane

Klasyfikacja	Synonimy
Grupa fenomenów	K+-aggravated myotonia K+-aggravated myotonia K-aggravated myotonia PAM K-aggravated myotonia PAM

Kod ORPHA	Kod OMIM	Kod ICD10
612	608390	G71.1

Kod ICD11

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*Źródło

orphanet