

Opis choroby *

Definicja

A rare ARX-related epileptic encephalopathy characterized by infantile onset of myoclonic epilepsy with generalized spasticity, severe global developmental delay, and moderate to profound intellectual disability. Obligate female carriers show subtle, generalized hyperreflexia. Late onset progressive spastic ataxia has also been reported.

Dane

Klasyfikacja

Choroba

Kod ORPHA

3175

Kod OMIM

308350

Kod ICD10

G25.3

Kod ICD11

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*Źródło

orphanet