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