Opis choroby *

Definicja

A rare genetic developmental and epileptic encephalopathy (DEE) characterized by developmental delay, generalized epilepsy consisting of eyelid myoclonia with absences and myoclonic-atonic seizures, intellectual disability and autism spectrum disorder (ASD).

Dane

Klasyfikacja
Choroba

Synonimy
SYNGAP1-related DEE
SYNGAP1-related DEE

Kod ORPHA
544254

Kod OMIM
-

Kod ICD10
G40.4

Kod ICD11
-

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*Źródło

orphanet