

## 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

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#### Kod ICD10

G40.4

#### Kod ICD11

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#### \*Źródło

orphanet