

## Opis choroby \*

### Definicja

A monogenic disease with epilepsy characterized by developmental delay and infantile spasms in the first months of life, followed by chorea and generalized dystonia and progressing to quadriplegic dyskinesia, recurrent status dystonicus, intractable focal epilepsy and severe intellectual disability.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

364063

#### Kod OMIM

308350

#### Kod ICD10

G40.4

#### Kod ICD11

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### \*Źródło

orphanet