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