Opis choroby *

Definicja

A rare genetic neurological disorder characterized by infantile hypotonia, congenital ophthalmic anomalies (including strabismus, esotropia, nystagmus, and central visual impairment), global developmental delay and intellectual disability, behavioral abnormalities, and movement disorder (such as dystonia, chorea, hyperkinesia, stereotypies). Mild facial dysmorphism and skeletal deformities have also been reported. EEG testing shows marked abnormalities in the absence of overt epileptic seizures.

Dane

Klasyfikacja
Choroba

Synonimy
SYT1-related neurodevelopmental disorder
Zaburzenie neurorozwojowe związane z SYT1

Kod ORPHA
522077

Kod OMIM
618218

Kod ICD10
F84.8

Kod ICD11
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*Źródło

orphanet