Opis choroby *

Definicja

A rare genetic neurological disorder characterized by early onset of microcephaly, severe global developmental delay and cognitive impairment, dyskinesia and hyperkinetic movements, visual impairment, autistic behavior, stereotypies, sleep disturbance, epilepsy, and cerebral malformations (such as corpus callosum hypogenesis, forebrain anomaly, and delayed myelination). Speech is minimal or absent, and ambulation is not attained. Patients with a larger 14q12 microdeletion show a more severe phenotype than those with intragenic alterations, with the addition of facial dysmorphism and agenesis of the corpus callosum.

Dane

Klasyfikacja
Choroba

Synonimy
FOXG1-related epileptic-dyskinetic encephalopathy
Encefalopatia padaczkowa związana z FOXG1

Kod ORPHA
561854

Kod OMIM
-

Kod ICD10
Q04.8

Kod ICD11
-

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*Źródło

orphanet