## 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 Synonimy

Choroba FOXG1-related epileptic-dyskinetic

encephalopathy

Encefalopatia padaczkowa związana z FOXG1

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 561854
 Q04.8

**Kod ICD11** 

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## <u>\*Źródło</u>

orphanet