Opis choroby *

Definicja

A rare genetic syndromic intellectual disability characterized by developmental delay, intellectual disability, ataxia, and, more variably, seizures and short stature. Behavioral abnormalities may also be observed, as well as variable facial and other dysmorphic features (such as broad nasal bridge, hypertelorism, almond-shaped eyes, high-arched palate, and anomalies of the fingers and toes). Brain imaging may reveal dilated ventricles, small corpus callosum, or posterior fossa abnormalities.

Dane

Klasyfikacja
Choroba

Synonimy
PADDAS syndrome
Zespół PADDAS
Zespół niepełnosprawności rozwojowej, ataksji i drgawek związany z PUM1
SCA47
Spinocerebellar ataxia type 47

Kod ORPHA
589515

Kod OMIM
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Kod ICD10
G11.8

Kod ICD11
-

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*Źródło

orphanet