Opis choroby *

Definicja

Early-onset epileptic encephalopathy and intellectual disability due to GRIN2A mutation is a rare intellectual disability and epilepsy syndrome characterized by global developmental delay and mild to profound intellectual disability, multiple types of usually intractable focal and generalized seizures with variable abnormal EEG findings, and bilateral progressive parenchymal volume loss and thin corpus callosum on brain MRI.

Dane

Klasyfikacja
Choroba

Kod ORPHA
289266

Kod OMIM
245570

Kod ICD10
E72.1

Kod ICD11
8A61.2Y

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*Źródło

orphanet