

Opis choroby *

Definicja

A rare, genetic, epilepsy syndrome characterized by congenital alopecia, early-onset epilepsy, intellectual disability and speech delay. Large stature, delayed bone development and abnormal electroencephalogram have also been associated.

Dane

Klasyfikacja

Synonimy

Zespół wad wrodzonych Alopecia-epilepsy-intellectual disability syndrome, Moynahan type
Zespół łysienie-padaczka-oligofrenia, typu Moynahana

Kod ORPHA

2574

Kod OMIM

203600

Kod ICD10

G40.4

Kod ICD11

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*Źródło

orphanet