

Opis choroby *

Definicja

Ravine syndrome is an extremely rare genetic neurological disorder, reported in a small number of patients in a specific community on Reunion Island (Ravine region), characterized by infantile anorexia with irrepressible and repeated vomiting, acute brainstem dysfunction, severe failure to thrive, and progressive encephalopathy with MRI showing vanishing of medulla oblongata and cerebellar white matter and severe atrophy of pons, along with supra-tentorial periventricular white-matter hyperintensities and basal ganglia anomalies.

Dane

Klasyfikacja

Choroba

Synonimy

Progressive encephalopathy with severe infantile anorexia

Postępująca encefalopatia z ciężką anoreksją niemowlęcą

Zespół wyspy Reunion, anoreksji, niepohamowanych wymiotów i objawów neurologicznych

Reunion island-anorexia-vomiting which is irrepressible-neurological signs syndrome

Kod ORPHA

99852

Kod OMIM

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Kod ICD10

E75.2

Kod ICD11

8A44.3

*Źródło

orphanet