Opis choroby *

Definicja

Moyamoya disease with early-onset achalasia is an exceedingly rare autosomal recessive neurological disorder reported only in a few families so far. It is characterized by the association of early onset achalasia (manifesting in infancy) with severe intracranial angiopathy that is consistent with moyamoya angiopathy in most cases (moyamoya disease; see this term). Other variable associated manifestations include hypertension, Raynaud phenomenon, and livedo reticularis.

Dane

Klasyfikacja

Choroba

Kod ORPHA 401945

Kod OMIM 615750

Kod ICD10

Kod ICD11

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*Źródło

orphanet