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
I67.5

Kod ICD11
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*Źródło

orphanet