

## 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