

Opis choroby *

Definicja

A rare, genetic, epilepsy syndrome characterized by epilepsy, palpebral conjunctival telangiectasias, borderline to moderate intellectual disability, diminished serum IgA levels, shortened fifth fingers and dysmorphic facial features (including frontal hirsutism, synophrys, anteverted nostrils, prominent ears, long philtrum, irregular teeth implantation, micrognathia). No new cases have been described in the literature since 1978.

Dane

Klasyfikacja

Choroba

Kod ORPHA

1951

Kod OMIM

226850

Kod ICD10

G40.8

Kod ICD11

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*Źródło

orphanet