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