Opis choroby *

Definicja

PENS syndrome is a rare, genetic, neurocutaneous syndrome characterized by the presence of randomly distributed, small, white to yellowish, multiple, rounded or irregular polycyclically-shaped, epidermal keratotic papules and plaques of ''gem-like'' appearance with a rough surface, typically located on the trunk and proximal limbs, associated with variable neurological abnormalities, including psychomotor delay, epilepsy, speech and language impairment and attention deficit-hyperactivity disorder. Clumsiness, dyslexia and oftalmological abnormalities have also been reported.

Dane

Klasyfikacja
Choroba

Synonimy
Papular epidermal nevi with skyline basal cell layers syndrome
Zespół grudkowego znamienia naskórkowego z "skyline" warstwą komórek podstawnych

Kod ORPHA
313936

Kod OMIM
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Kod ICD10
Q82.5

Kod ICD11
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*Źródło

orphanet