

## 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