Opis choroby *

Definicja

X-linked reticulate pigmentary disorder is an extremely rare skin disease described in only four families to date and characterized in males by diffuse reticulate brown hyperpigmentated skin lesions developing in early childhood and a variety of systemic manifestations (recurrent pneumonia, corneal opacification, gastrointestinal inflammation, urethral stricture, failure to thrive, hypohidrosis, digital clubbing, and unruly hair and flared eyebrows), while in females, there is only cutaneous involvement with the development in early childhood of localized brown hyperpigmented skin lesions following the lines of Blaschko. This disease was first considered as a cutaneous amyloidosis, but amyloid deposits are an inconstant feature.

Dane

Klasyfikacja
Choroba

Synonimy
Familial cutaneous amyloidosis
Amyloidoza skórna sprzężona z chromosomem X
Choroba Partingtona
PDR
Rodzinna amyloidoza skórna
XLPDR
PDR
Partington disease
X-linked cutaneous amyloidosis
XLPDR

Kod ORPHA
85453

Kod OMIM
301220

Kod ICD10
L99.0*

Kod ICD11
5D00.Y

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*Źródło

orphanet