

# Siateczkowate zaburzenie barwnikowe sprzężone z chromosomem X

**Kod Orpha: 85453 Kod OMIM: 301220**

## 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 hyperpigmented 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](#)

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## **Rozszerzony opis choroby**

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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