

## Opis choroby \*

### Definicja

A rare, genetic, immune deficiency with skin involvement characterized by clinical triad of non-scarring alopecia affecting mainly the scalp, well-demarcated mucosal erythema and psoriasiform erythematous intertriginous plaques. Follicular keratosis, keratoconjunctivitis, cataracts, angular cheilitis, fissured tongue, and recurrent infections are additional clinical features. Histopathology of mucosal lesions show characteristic findings of dyskeratotic keratinocytes, vacuolated basal cells, lack of epithelial maturation and decreased number of desmosomes.

### Dane

#### Klasyfikacja

Zespół wad wrodzonych Urban-Schosser-Spohn syndrome  
Zespół Urbana, Schossera i Spohna

#### Synonimy

#### Kod ORPHA

1839

#### Kod OMIM

158310

#### Kod ICD10

K13.7

#### Kod ICD11

DA02.0

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#### \*Źródło

orphanet