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, keratoconjuctivitis, 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

Synonimy
Urban-Schosser-Spohn syndrome
Zespół Urbana, Schossera i Spohna

Kod ORPHA
1839

Kod OMIM
158310

Kod ICD10
K13.7

Kod ICD11
DA02.0

---

*Źródło

orphanet