

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

Kod ORPHA	Kod OMIM	Kod ICD10
1839	158310	K13.7

Kod ICD11
DA02.0

*Źródło

orphanet