Opis choroby *

Definicja

A rare ectodermal dysplasia syndrome characterized by the association of ectodermal dysplasia (with hypotrichosis affecting scalp hair, eyebrows, and eyelashes, and partial anodontia), ectrodactyly, and macular dystrophy (appearing as a central geographic atrophy of the retinal pigment epithelium and choriocapillary layer of the macular area with coarse hyperpigmentations and sparing of the larger choroidal vessels). Variable additional limb defects (including absence deformities, polydactyly, syndactyly, or camptodactyly) have also been described, the hands often being more severely affected than the feet.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Ectodermal dysplasia-ectrodactyly-macular dystrophy syndrome
Dysplazja ektodermalna - ektrodaktylia - dystrofia plamki

Kod ORPHA
1897

Kod OMIM
225280

Kod ICD10
Q87.8

Kod ICD11
LD27.0Y

---

*Źródło

orphanet