

## 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

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych	Ectodermal dysplasia-ectrodactyly-macular dystrophy syndrome Dysplazja ektodermalna - ektrodaktylia - dystrofia plamki

<b>Kod ORPHA</b> 1897	<b>Kod OMIM</b> 225280	<b>Kod ICD10</b> Q87.8
<b>Kod ICD11</b> LD27.0Y		

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### \*Źródło

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