

# Dysplazja ektodermalna - ślepotą

## Kod Orpha: 1806 Kod OMIM: 268320

### Opis choroby \*

#### Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by intellectual disability, severe visual impairment due to ocular malformations (microphthalmos and microcornea with sclerocornea), short stature, hypotrichosis, dental anomalies, and dysmorphic facial features (such as a narrow nasal bridge with marked distal flaring and low-set, protruding ears). There have been no further descriptions in the literature since 1992.

#### Dane

##### Klasyfikacja

Zespół wad  
wrodzonych

**Kod ORPHA**  
1806

**Kod OMIM**  
268320

**Kod ICD10**  
Q87.8

**Kod ICD11**  
LD27.0Y

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\*Źródło

orphanet

### Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.