

Opis choroby *

Definicja

X-linked corneal dermoid (X-CND) is an exceedingly rare, benign, congenital, corneal tumor characterized by bilateral opacification of the cornea with superficial grayish layers and irregular raised whitish plaques, as well as fine blood vessels covering the central cornea, and intact peripheral corneal borders. No other ocular or systemic abnormality is noted. The pattern of inheritance described in the affected family is consistent with X-linked transmission.

Dane

Klasyfikacja

Choroba

Synonimy

Corneal dystrophy epithelial-short stature syndrome

Nabłonkowa dystrofia rogówki - niski wzrost

Zespół Guízar-Vázquez i Luengas-Muñoza

Guízar Vázquez-Luengas-Muñoz syndrome

Kod ORPHA

1661

Kod OMIM

304730

Kod ICD10

Q13.4

Kod ICD11

2F36.Y

*Źródło

orphanet