

Opis choroby *

Definicja

A rare, syndromic nevus characterized by the association of typically unilateral, closely arranged, linear, slightly elevated, multiple, nevus comedonicus lesions located usually on the face, neck, trunk or limbs (with or without a central, dark, firm, hyperkeratotic plug and secondary acneiform lesions) with extracutaneous ocular, skeletal, and/or central nervous system abnormalities, such as ipsilateral cataract, corneal erosion, poly-/syndactyly, absent fifth finger, scoliosis, vertebral defects, corpus callosum agenesis, seizures, interhemispheric cyst, intellectual deficiency, and/or developmental delay.

Dane

Klasyfikacja

Choroba

Kod ORPHA

64754

Kod OMIM

617025

Kod ICD10

Q82.5

Kod ICD11

LD2D.Y

*Źródło

orphonet