Opis choroby *

Definicja

A rare genetic syndromic intellectual disability characterized by language delay and mild to moderate intellectual disability associated with truncal obesity, congenital nonprogressive retinal dystrophy with poor night vision and reduced visual acuity, and micropenis in males. Cataracts may occur in the second or third decade of life.

Dane

Klasyfikacja
Choroba

Synonimy
Intellectual disability-truncal obesity-retinal dystrophy-micropenis syndrome
Niepełnosprawność intelektualna - otyłość - dystrofia siatkówki - mikropenis

Kod ORPHA
75858

Kod OMIM
610156

Kod ICD10
Q87.8

Kod ICD11
9B70

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*Źródło

orphanet