

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

*Źródło

orphanet