Opis choroby *

Definicja

A rare, genetic, primary lipodystrophy syndrome characterized by severe developmental delay and intellectual disability, hypertonia, hyperreflexia, microcephaly, tightly adherent skin, an aged appearance, severe generalized lipodystrophy, and distinct facial dysmorphism which includes large prominent eyes, narrow nasal bridge, tented upper lip vermilion, an open mouth, and high-arched palate. Laboratory analysis of serum and urine are normal.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Generalized lipodystrophy-progeroid features-severe intellectual disability syndrome
Zespół lipodystrofii uogólnionej, cech progeroidalnych i ciężkiej niepełnosprawności intelektualnej

Kod ORPHA
435628

Kod OMIM
614098

Kod ICD10
E88.1

Kod ICD11
-

---

*Źródło

orphanet