Opis choroby *

Definicja

A rare genetic disease characterized by severe pre- and postnatal growth failure with short stature and microcephaly, facial dysmorphism (including a small jaw and prominent midface), severe insulin resistance, fatty liver, and hypertriglyceridemia developing in childhood, and primary gonadal failure. Mild global learning difficulties and acanthosis nigricans have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
436182

Kod OMIM
616541

Kod ICD10
Q87.1

Kod ICD11
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*Źródło

orphanet