

Opis choroby *

Definicja

A rare genetic lipodystrophy characterized by loss of subcutaneous adipose tissue primarily affecting the lower limbs and gluteal region due to a defect in the PLIN1 gene. Associated features of insulin resistance, hepatic steatosis, dyslipidemia, hypertension, axillary acanthosis nigricans and muscular hypertrophy of the lower limbs are typical.

Dane

Klasyfikacja

Choroba

Synonimy

FPLD4
FplD zależna od plIN1
FplD4
PLIN1-related FPLD

Kod ORPHA

280356

Kod OMIM

613877

Kod ICD10

E88.1

Kod ICD11

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*Źródło

orphanet