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