Opis choroby *

Definicja

A rare, genetic lipodystrophy characterized by abnormal subcutaneous fat distribution, resulting in excess accumulation of fat in the face, neck, shoulders, axillae, trunk and pubic region, and loss of subcutaneous fat from the lower extremities. Variable common additional features are progressive adult onset myopathy, insulin resistance, diabetes, hypertriglyceridemia, hepatic steatosis, and vitiligo.

Dane

Klasyfikacja
Choroba

Synonimy
FPLD6
FplD związana z LIPE
FplD6
LIPE-related FPLD

Kod ORPHA
435660

Kod OMIM
615980

Kod ICD10
E88.1

Kod ICD11
5A44

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*Źródło

orphanet