PLIN1-related familial partial lipodystrophy

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. PLIN1-related familial partial lipodystrophy. ORPHA:280356

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.