

What is Lipodystrophy?

Lipodystrophy related diseases are a heterogeneous group of rare diseases characterized by generalized or partial lack of subcutaneous adipose tissue which can be genetic or acquired. Lack or significantly decreased amounts of adipose tissue in patients results in dyslipidemia and insulin resistance and many other metabolic and cardiac comorbidities, associated with high incidence of mortality.

Why Study Lipodystrophy?

Although lipodystrophy-related diseases are rare, they serve as an exemplary model to study and understand the pathogenesis of metabolic diseases in adipose disorders such as obesity. In turn, this can lead to new therapeutic targets to treat and/or prevent metabolic disease(s).

Rare Is Everywhere

- Lipodystrophy is one of 7,000+ rare diseases in the U.S. identified by the National Institutes of Health.
- Rare diseases affect 1 in 10 people in the U.S., that's 30 million people.
- 95% of rare diseases have no FDA approved treatment.
- About 50% of rare diseases do not have a disease-specific organization researching or supporting the condition.

LipodystrophyUnited.org

Visual Symptoms can include some, or all of the following:

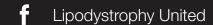
- Striking fat loss in some parts or all of the body
- Fat accumulation in areas such as the upper back or neck and labia majora (females)
- Very muscular appearance (muscular pseudohypertrophy)
- Prominent veins
- Dark, thickened patches of skin around the creases of neck, arms, groin or other areas (acanthosis nigricans due to severe insulin resistance)
- Itchy bumps in the hands, feet, arms, legs and bottom (xanthomas)

Non-Visual Symptoms can include some, or all of the following:

- Cardiac problems, including cardiovascular disease, cardiomyopathy and conduction abnormalities
- Polycystic Ovarian Syndrome (PCOS) (often displaying as dysmenorrhea, enlarged ovarian mass (with or without active ovarian follicles or cysts), hirsutism, female pattern hair loss)
- Dyslipidemia (elevated serum triglycerides, cholesterol, low HDL/high LDL)
- Uncontrollable diabetes, resistance to conventional insulin sensitizing therapies
- Severe insulin resistance with increased insulin therapy requirements
- Fatty Liver (palpable) and/or Non-Alcoholic Steatohepatitisis (NASH)
- Hyperphagia (increased hunger)
- Increased incidence of pancreatitis
- Nephropathies (most commonly proteinurea)
- Gastroparesis and/or idiopathic irritable bowel syndrome
- Generalized or localized muscle pain or inflammation
- Hypertension







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