

TREATMENT AND MANAGEMENT

Disease-specific:

- Metreleptin (Mylept) - FDA-approved leptin replacement for generalized lipodystrophy and other investigational drugs in clinical studies.

Standards of care medications for associated comorbidities:

- Insulin, metformin, SGLT-2 inhibitors, GLP-1/GIP RAs, thiazolidinediones, or other approved drugs for diabetes and metabolic control.
- Fibrates + omega-3 fatty acids for hypertriglyceridemia
- Liver, kidney, and heart monitoring
- Dietary guidance and specialist nutrition support
- Psychological support for mental health and body image



DIFFERENTIAL DIAGNOSIS

Lipodystrophy is frequently misdiagnosed due to phenotypic overlap with common metabolic disorders - clinicians should actively consider it as a potential underlying diagnosis, not merely rule it out:

- Type 2 diabetes mellitus / metabolic syndrome
- Obesity / common overweight
- Polyendocrine Metabolic Ovarian Syndrome (PMOS) as a stand-alone diagnosis.
- Cushing's syndrome
- Familial hypertriglyceridemia

CONTACT US!

✉ Info@lipodystrophyunited.org

📷 [LipodystrophyUnited](#)

✂ [LipodystrophyLU](#)

🌐 [Lipodystrophy United](#)

📘 [Lipodystrophy United](#)

🦋 [lipodystrophyunite.bsky.social](#)

22 Frances Rd, Los Lunas NM 87031



Website



Donate



COULD YOUR PATIENT HAVE LIPODYSTROPHY?





HOW TO RECOGNIZE LIPODYSTROPHY

How to recognize lipodystrophy:

1. Abnormal fat distribution

Generalized or partial loss of subcutaneous fat, often with prominent muscles and veins in the limb, acromegaloid or cushingoid appearance. Might present with fat sparing or excess in the face, neck, or abdomen.

2. Insulin resistance:

Diabetes requiring high-dose insulin treatment; acanthosis nigricans, PCOS or oligomenorrhoea in women.

3. Dyslipidemia:

hypertriglyceridemia, low HDL-cholesterol, history of acute pancreatitis.

4. Hepatic steatosis:

Findings consistent with Metabolic Dysfunction-Associated Steatotic Liver Disease (MASLD) in the absence of other identifiable causes.

5. Cardiovascular complications:

Premature atherosclerosis, hypertrophic cardiomyopathy, arrhythmias and conduction defects



DIAGNOSIS

Diagnostic Approach:

Diagnosis proceeds through two sequential phases: clinical recognition followed by subtype characterization.

Phase 1 - Clinical Assessment:

- Medical history, family history, and physical examination
- Laboratory evaluation (fasting lipids, glucose, insulin, HbA1c, liver function)
- Body composition and liver imaging
- Establishing the clinical diagnosis

Phase 2 - Subtype Characterization:

- Genetic Testing: NGS gene panel covering established lipodystrophy loci is the recommended first-line investigation. WES/WGS may be indicated when panel testing is uninformative.
- Immunological Evaluation: Essential when suspecting acquired types. Associated with autoimmune hepatitis, dermatomyositis, panniculitis and other autoimmune conditions. Some types associated with low C3 complement.



WHAT IS LIPODYSTROPHY?

Lipodystrophy syndromes comprise a heterogeneous group of rare genetic or acquired disorders characterized by selective loss, deficiency, and/or dysfunction of adipose tissue, which may be generalized or partial. Impaired adipose tissue storage capacity leads to ectopic lipid deposition in non-adipose organs, resulting in progressive and often severe metabolic complications, including insulin resistance, dyslipidemia, hepatic steatosis, and cardiovascular comorbidities, with consequent increased risks of morbidity and premature mortality.