

# PATIENT'S GUIDE TO LIPODYSTROPHY



## What is Lipodystrophy?

Lipodystrophy is a rare disease in which people do not have enough fat in some or all of their body.

Lipodystrophy is progressive and can lead to serious and life-threatening complications.

## How do you diagnose lipodystrophy?

Lipodystrophy is a clinical diagnosis typically confirmed by an endocrinologist using a skinfold caliper or DEXA scan, in combination with blood tests for triglycerides, glucose levels, insulin resistance and sometimes leptin levels. While most forms of lipodystrophy are genetic, some can be acquired. Recent research has led to an increase in identifications of mutations, but not all mutations have been identified so a genetic test can confirm a diagnosis of some types of genetic lipodystrophy but not all types.

## Why seek diagnosis?

Without proper fat distribution, the body can have difficulty storing fats and sugars from food. The fat (triglycerides) and sugar (glucose) remain in the blood and can store in the organs leading to problems with organ function such as liver, kidney and heart disease. Early diagnosis can change treatment options and potentially slow the progression of the disease.

## 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.



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

- Very little fat in some or all parts of the body
- More fat in some or all parts of the body
- Very muscular appearance where there is no fat
- Very visible or large appearance of veins under the skin
- Itchy and painful bumps in the hands, feet, arms, legs and bottom (xanthomas)
- Dark patches of skin around the creases of neck, armpits, groin or other areas (acanthosis nigricans due to severe insulin resistance)

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

- Heart problems such as cardiovascular disease, cardiomyopathy or abnormal heart rhythm
- Polycystic Ovarian Syndrome or absence of menses depending on subtype of lipodystrophy
- Fatty Liver or Non-Alcoholic Steatosis (NASH)
- High triglycerides
- Difficult to treat diabetes
- Severe insulin resistance
- High Blood Pressure
- Extreme hunger
- Pancreatitis
- Kidney Problems
- Gastroparesis
- Pain in muscle or joints



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