CONDITION

Lipodystrophy

Lipodystrophy is a group of conditions that cause abnormal loss or distribution of body fat and may lead to serious metabolic problems.

WHAT IT IS
Abnormal loss or distribution of body fat

BODY SYSTEM
Body fat, hormones, liver, heart and circulation

MAIN SYMPTOMS
Visible fat loss, fat buildup, strong hunger, muscle prominence

WHAT CAUSES IT
Inherited changes, immune conditions, medicines, or unknown causes

HOW IT IS DIAGNOSED
Examination, metabolic tests, imaging, and sometimes genetic testing

MAIN TREATMENT
Metabolic care, nutrition support, and selected hormone therapy

CAN IT BE CURED?
Sometimes - acquired forms may improve after trigger removal

IS IT AN EMERGENCY?
Sometimes - severe abdominal pain may mean pancreatitis

LONG-TERM OUTLOOK
Often lifelong, with outlook shaped by metabolic complications

Overview

Lipodystrophy is a group of conditions in which the body has too little fat in certain places or stores fat in an unusual pattern. It can affect nearly the whole body, called generalized lipodystrophy, or specific areas, called partial or localized lipodystrophy.

Body fat is also called adipose tissue. It does more than shape the body or store energy. It produces hormones, including leptin, and helps keep excess fat out of the liver, muscles, and bloodstream.

When healthy fat tissue is missing, the body may store fat in organs where it does not belong. This can cause severe insulin resistance, meaning insulin has difficulty controlling blood sugar. High triglycerides, fatty liver, diabetes, and inflammation of the pancreas can follow.

The visible changes are not the same for everyone. Some people look very muscular because there is little fat beneath their skin. Others lose fat from their arms and legs but gain it around the neck, face, or abdomen. These changes are not simply caused by diet or exercise.

Symptoms

Symptoms depend on which areas are affected and whether the condition began during childhood or adulthood.

Possible physical changes include:

Loss of body fat may make hands, feet, or facial features appear larger or more prominent. Children with generalized forms may grow quickly, have a strong appetite, and appear muscular from an early age.

Metabolic problems may not cause symptoms at first. When symptoms occur, they can include intense thirst, frequent urination, tiredness, blurred vision, or repeated infections from high blood sugar.

Very high triglycerides can cause pancreatitis, which is inflammation of the pancreas. This usually causes severe, persistent upper abdominal pain, often with nausea or vomiting.

Some people develop an enlarged liver. Menstrual changes, excess facial or body hair, acne, and fertility problems may occur, especially when insulin resistance affects hormone balance.

Localized lipodystrophy may appear as a small dent or lump at an injection site. It often does not cause the widespread metabolic problems seen in generalized or partial forms.

- Very little fat beneath the skin
- Arms or legs that look unusually muscular or defined
- Prominent veins
- Loss of fat from the face, buttocks, arms, or legs
- Fat buildup around the neck, chin, face, or abdomen
- Dark, thickened, velvety skin, especially around the neck or armpits

Causes and risk factors

Lipodystrophy may be inherited or acquired later in life.

Inherited forms are caused by changes in genes involved in the development, storage, or breakdown of fat. Some begin in infancy. Others, including several familial partial forms,

become clearer around puberty or early adulthood. A person can carry a gene change even when other relatives have mild or unrecognized symptoms.

Acquired forms are not caused by an inherited gene change. They may be associated with:

HIV-associated lipodystrophy can involve fat loss, central fat buildup, or both. HIV itself, past treatment, current treatment, age, and other metabolic factors may all contribute. HIV medicine should not be stopped or changed without specialist advice.

Repeated insulin or other injections can cause local fat loss or a fatty lump. Using the same damaged area again may make medicine absorption less predictable.

Having an inherited form is not related to personal choices, body weight, or a lack of exercise.

- An immune system attack on fat tissue
- Another autoimmune condition
- Certain infections or medicines
- Repeated injections into the same area
- Previous HIV treatment, particularly some older medicines
- An unknown trigger

Diagnosis

There is no single test that confirms every form of lipodystrophy. Diagnosis usually begins with a detailed medical and family history and an examination of where fat is present or missing.

Photographs taken over time can help show whether body shape has changed. Skinfold measurements or scans, such as dual-energy X-ray absorptiometry or MRI, may be used to assess fat distribution. These scans support the diagnosis but do not replace an experienced clinical assessment.

Blood tests may check:

Leptin levels may be low, especially in generalized lipodystrophy, but a leptin result alone cannot confirm or rule out the condition.

Genetic testing can confirm some inherited forms. A negative result does not exclude lipodystrophy because not every responsible gene change is known or detected by current tests.

Further tests may look for fatty liver, pancreatitis, heart problems, or signs of an autoimmune condition. The assessment also helps distinguish lipodystrophy from ordinary weight loss, malnutrition, muscle conditions, and other causes of altered body shape.

- Blood glucose and long-term glucose control
- Triglycerides and cholesterol
- Liver function
- Kidney function
- Reproductive hormones when relevant

Treatment and management

Treatment aims to control metabolic complications, support nutrition, and address physical or emotional effects. The plan differs according to the type of lipodystrophy and the organs affected.

Food choices are important, but severe fat loss cannot usually be reversed by eating more. Nutrition support often focuses on meeting growth and energy needs while limiting sharp rises in blood sugar and triglycerides. Extreme food restriction can be harmful, especially for children or people with very low body fat.

Regular physical activity can improve blood sugar, triglycerides, and heart health. The type and intensity should take account of heart, liver, eye, or muscle complications.

Diabetes may require glucose-lowering medicines or insulin. Very high insulin doses are sometimes needed because insulin resistance can be severe. Treatment may also be needed for high triglycerides, high blood pressure, fatty liver, or kidney problems.

Metreleptin is a manufactured form of the hormone leptin. In the United States, it is approved as an addition to diet for metabolic complications of generalized lipodystrophy. Approval for partial lipodystrophy differs between countries. It is not a cosmetic treatment or a general weight-loss medicine. It requires specialist supervision because it has important risks and monitoring requirements.

If a medicine or repeated injection has caused localized changes, adjusting the injection technique or changing the site may prevent further damage. Prescribed treatment should not be stopped without advice.

For HIV-associated changes, an HIV specialist may review the treatment regimen and metabolic risks. Changing HIV medicine does not always reverse established fat loss.

Fillers, fat transfer, or reconstructive surgery may help selected people with facial or body changes. Results can be limited if fat loss is still progressing. Psychological support may also help with distress, stigma, or changes in self-image.

Complications

The main health risks come from fat being stored in the liver, muscles, and bloodstream rather than in healthy fat tissue.

Possible complications include:

Not everyone develops all of these complications. Risk is often greater when fat loss is widespread, but some partial forms can also cause severe metabolic problems.

Changes in appearance may affect relationships, school, work, and mental well-being. These effects are part of the condition and deserve attention alongside physical health.

- Severe insulin resistance and diabetes
- Very high triglycerides
- Fatty liver, inflammation, scarring, or cirrhosis
- Pancreatitis
- Heart and blood vessel problems
- Kidney damage related to diabetes or the underlying condition
- Menstrual, fertility, and other hormone problems

Prevention

Inherited lipodystrophy cannot currently be prevented. Genetic counselling can explain how a diagnosed form may be passed through a family and discuss testing options.

Most autoimmune or unexplained acquired forms also cannot be reliably prevented.

Localized changes from injections may sometimes be reduced by rotating injection sites, using the correct technique, and avoiding repeated injections into dents, scars, or fatty lumps. HIV treatment should be chosen and adjusted by an HIV specialist rather than changed to prevent body-shape changes without considering viral control.

Good metabolic care does not prevent the underlying fat disorder, but it may reduce complications involving the liver, pancreas, heart, and blood vessels.

Living with it and what to expect

Many inherited forms are lifelong. The pattern of fat loss may remain fairly stable, or it may

become more noticeable during growth, puberty, pregnancy, or ageing. Acquired forms may progress for a time and then stabilize. Some localized forms can improve after the trigger is removed.

Metabolic problems can change even when body shape looks unchanged. Blood sugar, triglycerides, liver health, blood pressure, and kidney function therefore need routine follow-up. Children may also need growth and puberty monitored.

With ongoing care, many people study, work, exercise, have relationships, and raise families. Daily life may still involve food planning, medicines, blood tests, injections, or specialist visits. The long-term outlook is influenced more by the severity and control of metabolic complications than by appearance alone.

Routine follow-up is also a chance to discuss pain, fertility, pregnancy planning, emotional well-being, or unwanted attention related to physical changes.

When to get medical help

Arrange a routine medical appointment if you notice unexplained loss of fat, an unusual pattern of fat buildup, increasing muscle definition without a clear reason, or persistent dents and lumps at injection sites. Earlier assessment is especially useful when similar changes occur in relatives or begin during childhood.

Seek prompt medical advice for symptoms of high blood sugar, such as increasing thirst, frequent urination, blurred vision, unexplained weight loss, or repeated infections. Yellow skin or eyes, a swollen abdomen, or worsening fatigue also needs assessment.

Get urgent or emergency help for:

These symptoms can have several causes. In someone with lipodystrophy, they may indicate pancreatitis, a serious blood sugar problem, or a heart or circulation emergency.

- Severe or persistent upper abdominal pain, especially with vomiting
- Rapid breathing, marked drowsiness, confusion, or inability to keep fluids down
- Chest pressure, sudden weakness, facial drooping, or difficulty speaking
- Fainting or severe shortness of breath

Common questions

Is lipodystrophy the same as being very thin?

No. A thin person may still have a normal distribution and function of body fat. Lipodystrophy causes selective or widespread loss of fat tissue and may lead to fat collecting in the liver, muscles, or blood.

Can lipodystrophy cause diabetes?

Yes. Missing or poorly functioning fat tissue can make the body highly resistant to insulin. This increases the risk of diabetes, but not everyone with lipodystrophy develops it.

Can the lost fat grow back?

Sometimes. Localized or acquired fat loss may improve if the trigger is removed, although recovery can be incomplete. Inherited generalized and partial forms are usually long term. Metabolic treatment can improve health without restoring the missing fat.

Is lipodystrophy inherited?

Some forms are inherited, while others develop because of immune conditions, medicines, injections, infections, or unknown causes. Genetic testing and family history can help identify an inherited form.

Can lipodystrophy affect the face?

Yes. Fat loss may make the cheeks and temples look hollow or make facial features appear more prominent. Other forms cause fat to collect around the face, chin, or neck.

What is HIV-associated lipodystrophy?

It is a pattern of fat loss, fat buildup, or both in some people living with HIV. It has been linked particularly to older HIV treatments, although several factors may contribute. Established changes do not always reverse when treatment is changed.

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