CONDITION

Lymphangioleimyomatosis

LAM is a rare condition in which abnormal cells cause lung cysts and may affect the lymphatic system and kidneys.

WHAT IT IS
Rare condition causing lung cysts and abnormal LAM-cell growth

BODY SYSTEM
Lungs, lymphatic system, and kidneys

MAIN SYMPTOMS
Shortness of breath, collapsed lung, cough, chest discomfort

WHAT CAUSES IT
TSC1 or TSC2 gene changes, usually acquired

HOW IT IS DIAGNOSED
High-resolution chest CT plus VEGF-D testing or biopsy

MAIN TREATMENT
Sirolimus, breathing support, and complication-specific procedures

CAN IT BE CURED?
No, but treatment can slow progression

IS IT AN EMERGENCY?
Sometimes, with sudden chest pain or severe breathlessness

LONG-TERM OUTLOOK
Often slowly progressive; treatment can stabilize lung function

Overview

Lymphangioleiomyomatosis, usually shortened to LAM, is a rare condition that mainly affects the lungs. "Lymphangioleimyomatosis," used in this article's title, is a common misspelling of the medical name.

In LAM, abnormal muscle-like cells grow in the lungs and sometimes in the lymphatic system and kidneys. The lymphatic system is a network of vessels that carries fluid around the body and supports the immune system.

LAM cells can damage lung tissue and create many small, air-filled spaces called cysts. Over time, this can make it harder to move air out of the lungs. A cyst can also rupture, allowing air to collect around a lung and causing it to collapse. This is called a pneumothorax.

LAM occurs almost entirely in women, especially before menopause. It can develop on its own, called sporadic LAM, or in people with tuberous sclerosis complex, often shortened to TSC. TSC is a genetic condition that can cause noncancerous growths in several organs.

LAM is long-lasting, but its course is not the same for everyone. Some people have few symptoms and slow change. Others lose lung function more quickly. Treatment can slow decline, manage complications, and help many people remain active.

Symptoms

Shortness of breath is the most common symptom. It may first appear during exercise, climbing stairs, or walking uphill. As lung function changes, everyday activities may require more effort.

Other possible symptoms include:

LAM can block lymphatic vessels and cause chyle, a milky fluid containing fat, to collect around the lungs. This is called a chylous pleural effusion. It may cause increasing breathlessness, chest heaviness, or cough.

Some people have kidney growths called angiomyolipomas. These are usually noncancerous and contain blood vessels, muscle, and fat. Many cause no symptoms. Larger growths can bleed, causing pain in the side or back, blood in the urine, dizziness, or weakness.

Symptoms can resemble asthma, chronic obstructive pulmonary condition, or other lung problems. Having breathlessness or lung cysts does not by itself mean that you have LAM.

- A recurring dry cough or wheeze
- Chest discomfort
- Tiredness and reduced exercise tolerance
- Repeated collapsed lungs
- Swelling in the abdomen or legs from lymphatic fluid
- Coughing up blood, which is less common

Causes and risk factors

LAM cells usually have changes in one of two genes, TSC1 or TSC2 . These genes normally help control cell growth. When they stop working properly, a growth pathway known as mTOR becomes overactive.

In sporadic LAM, the gene change develops in certain cells during a person's life. It is not present throughout the body and is not usually passed to children.

In TSC-associated LAM, a person has an inherited or newly occurring TSC1 or TSC2 change throughout the body. TSC can run in families, although some people are the first in their family to have it.

The hormone estrogen appears to influence LAM-cell growth. This helps explain why LAM occurs mostly in women and why lung function may change during pregnancy or after exposure to estrogen-containing medicines. However, estrogen alone does not cause LAM.

Smoking does not cause LAM. It can add further lung damage and reduce the breathing reserve available to someone who already has the condition.

Diagnosis

Diagnosis usually begins with a high-resolution CT scan of the chest. This detailed scan can show the thin-walled lung cysts typical of LAM. A standard chest X-ray may miss these changes and cannot confirm the condition.

A CT pattern that looks like LAM is usually combined with other evidence. This may include:

VEGF-D is a protein involved in lymphatic vessel growth. A sufficiently raised level can support a diagnosis without a lung biopsy in the right clinical setting. A normal result does not completely rule out LAM.

If imaging and other tests do not provide enough evidence, a biopsy may be considered. A biopsy removes a small tissue sample so that it can be examined for LAM cells. Because biopsy has risks, it is not automatically needed for everyone with a characteristic CT scan.

Breathing tests measure how well air moves through the lungs and how efficiently oxygen crosses into the blood. They help establish a baseline and track the condition but do not confirm LAM by themselves.

Testing may also include kidney imaging, oxygen measurements, and an assessment for signs of TSC. Genetic counselling or testing may be appropriate if TSC is suspected.

- A raised blood level of vascular endothelial growth factor D, or VEGF-D
- Kidney angiomyolipomas
- Chylous fluid around the lungs or in the abdomen
- Lymphatic growths called lymphangioleiomyomas
- A confirmed diagnosis of tuberous sclerosis complex

Treatment and management

There is no treatment that removes all LAM cells, but treatment can slow lung-function decline and control complications.

Sirolimus is the main medicine used to treat LAM. It blocks the overactive mTOR growth pathway. It may be offered when lung function is abnormal or declining, and it can also help some lymphatic complications and angiomyolipomas. It generally controls the condition only while it is being taken.

Sirolimus requires regular blood tests and clinical monitoring. Possible effects include mouth sores, diarrhoea, ankle swelling, high blood fats, and a greater risk of some infections. Treatment decisions consider lung function, symptoms, other health conditions, and plans for pregnancy.

Other parts of care may include:

A collapsed lung usually needs prompt assessment. Because it often happens again in LAM, a procedure called pleurodesis may be recommended after the first episode. Pleurodesis helps the lung lining stick to the chest wall, reducing the chance of another collapse. Having this procedure does not automatically prevent future lung transplantation.

Estrogen treatment is not used to control LAM. Medicines containing estrogen, including some contraceptives and menopause treatments, need an individual discussion about risks and alternatives. Treatments once tried for LAM, such as removing the ovaries or using progesterone solely to treat the condition, have not shown reliable benefit.

- Inhaled bronchodilators if tests show reversible narrowing of the airways
- Pulmonary rehabilitation to build fitness and manage breathlessness
- Supplemental oxygen when blood oxygen is consistently low
- Procedures to drain or control chylous fluid
- Monitoring or treatment of kidney angiomyolipomas
- Lung transplantation for advanced breathing impairment

Complications

Pneumothorax is a major complication. It happens when air leaks from a lung cyst into the space between the lung and chest wall. The lung may partly or completely collapse. It can recur, sometimes on either side.

Other complications include chylous fluid around the lungs or in the abdomen, worsening airflow obstruction, low blood oxygen, and bleeding from kidney angiomyolipomas.

Advanced LAM can lead to respiratory failure, meaning the lungs cannot supply enough oxygen or remove carbon dioxide adequately. This usually develops gradually rather than without warning. Lung-function tests and oxygen checks help identify changes before they become severe.

Sirolimus can also cause complications, including infections and changes in blood counts, liver tests, or cholesterol. Regular monitoring is part of using it safely.

Prevention

LAM itself cannot currently be prevented. The cell changes responsible for sporadic LAM happen without a known preventable trigger. TSC-associated LAM results from a genetic change and is also not caused by lifestyle choices.

You can still protect the lung function you have. Avoiding smoking and second-hand smoke reduces added lung injury. Recommended respiratory vaccinations may lower the chance of preventable infections, although they do not prevent LAM.

If you have TSC and could develop LAM, planned screening may detect lung changes before symptoms become severe. Genetic counselling can explain how TSC may affect relatives or future children.

Living with it and what to expect

LAM is usually monitored over months and years with breathing tests, symptom reviews, oxygen measurements, and imaging when needed. Routine appointments help show whether lung function is stable or changing. A single test result rarely tells the whole story.

The rate of progression differs widely. Some people remain stable for long periods, while others have a steady decline or repeated complications. Lung-function loss often slows after menopause, but LAM does not necessarily stop progressing. Sirolimus can stabilize lung function for many people who need treatment, but it is not a cure.

Regular activity is generally encouraged within your breathing limits. Pulmonary rehabilitation can teach pacing, breathing techniques, and safe ways to improve fitness. Breathlessness does not always mean that lung damage has suddenly worsened, but a clear or lasting change should be assessed.

Pregnancy may increase the risk of a collapsed lung, chylous fluid, and reduced lung function. Sirolimus also requires specific planning around pregnancy. Anyone considering pregnancy should have a pre-pregnancy discussion with clinicians familiar with LAM and high-risk pregnancy care.

Air travel is possible for many people with stable LAM, but changing cabin pressure may increase pneumothorax risk. Do not fly with a current or recently treated collapsed lung until a qualified clinician has confirmed that flying is safe.

Many people continue working, travelling, exercising, and caring for families. Adjustments may become necessary if breathlessness progresses. Emotional support, practical planning, and contact with a LAM patient organisation can help with the uncertainty of a rare condition.

When to get medical help

Seek emergency medical care for:

These symptoms may indicate a collapsed lung, serious bleeding from a kidney angiomyolipoma, or another urgent problem.

Arrange prompt medical advice for a noticeable, persistent increase in breathlessness, new chest discomfort, swelling of the abdomen or legs, blood in the urine, or possible infection while taking sirolimus.

Routine follow-up is appropriate when symptoms are stable. Keep planned appointments for lung-function testing, medicine monitoring, oxygen assessment, and kidney imaging. Report gradual changes rather than waiting for the next visit if they begin to affect everyday activities.

- Sudden chest pain with new or rapidly worsening breathlessness
- Severe difficulty breathing, blue or grey lips, fainting, or confusion
- Sudden severe side or back pain with weakness, dizziness, or collapse
- Coughing up more than a small streak of blood

Common questions

Is LAM cancer?

LAM is not classified as a typical cancer. However, LAM cells can grow abnormally, travel through blood or lymphatic vessels, and appear in different parts of the body. This cancer-like behaviour is one reason long-term specialist monitoring is needed.

How fast does LAM progress?

There is no single rate. Lung function may remain stable for years or decline gradually, and complications such as pneumothorax can interrupt an otherwise stable period. Repeated breathing tests are more informative than one result.

Can LAM be cured?

There is currently no cure. Sirolimus can slow or stabilize progression while it is taken. Lung transplantation can replace severely affected lungs, but it is reserved for advanced impairment and involves lifelong follow-up.

Can I exercise with LAM?

Many people can and should remain active within safe limits. Walking, cycling, strength work, and supervised pulmonary rehabilitation may improve stamina and confidence. Stop and seek advice if activity causes sudden chest pain, severe breathlessness, dizziness, or fainting.

Can I get pregnant if I have LAM?

Pregnancy is possible, but it can increase the chance of lung and lymphatic complications. Risks are different for each person and are influenced by current lung function, previous pneumothorax, kidney angiomyolipomas, and treatment. Planning before pregnancy is important.

Is LAM inherited?

Sporadic LAM is not usually inherited. LAM associated with tuberous sclerosis complex can occur because of an inherited TSC1 or TSC2 gene change. Genetic counselling can clarify the implications when TSC is present or suspected.

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