CONDITION

Pulmonary lymphangiomyomatosis

Pulmonary lymphangiomyomatosis is a rare condition in which abnormal cells cause cysts and damage in the lungs.

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

BODY SYSTEM
Lungs, lymph vessels, and kidneys

MAIN SYMPTOMS
Breathlessness, cough, chest pain, collapsed lung

WHAT CAUSES IT
Gene changes affecting cell growth; sometimes tuberous sclerosis

HOW IT IS DIAGNOSED
High-resolution CT and VEGF-D testing; sometimes lung biopsy

MAIN TREATMENT
Sirolimus, symptom support, and sometimes lung transplantation

CAN IT BE CURED?
No - treatment can slow progression

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

LONG-TERM OUTLOOK
Often slowly progressive, with wide differences between people

Overview

Pulmonary lymphangiomyomatosis, usually shortened to LAM, is a rare condition that mainly affects the lungs. Abnormal muscle-like cells collect in and around the small airways, blood vessels, and lymph vessels. Over time, this can create many thin-walled air-filled spaces, called cysts, throughout the lungs.

These cysts can make it harder to move air in and out. They can also rupture, allowing air to leak around a lung and causing it to collapse. This is called a pneumothorax .

LAM can also block lymph vessels. Lymph is a clear fluid that carries immune cells and absorbed fats. A blockage may cause this fluid to collect around a lung, known as a chylous pleural effusion or chylothorax .

Some people with LAM develop non-cancerous kidney growths called angiomyolipomas . These contain blood vessels, muscle, and fat. Larger angiomyolipomas can sometimes bleed.

LAM occurs in two forms. Sporadic LAM develops on its own. TSC-LAM occurs in people with tuberous sclerosis complex , or TSC, an inherited condition that can cause growths in several organs. LAM affects almost entirely women, although it has rarely been reported in men, usually in connection with TSC.

Symptoms

Symptoms may begin gradually, and early LAM can be mistaken for asthma or another lung condition. Some people have few symptoms when lung cysts are first found.

Possible symptoms include:

A collapsed lung may be the first clear sign of LAM. It usually causes sudden one-sided chest pain and a rapid increase in breathlessness. Pneumothorax can happen more than once.

A chylous effusion may cause increasing breathlessness, chest heaviness, cough, or swelling of the abdomen. Rarely, a person may cough up blood.

Kidney angiomyolipomas often cause no symptoms. Bleeding may cause sudden pain in the side or back, blood in the urine, weakness, or dizziness.

- Shortness of breath, especially during activity
- Reduced ability to exercise
- A dry or occasional cough
- Wheezing
- Chest discomfort
- Tiredness

Causes and risk factors

LAM cells have changes in genes that normally control cell growth. These are commonly the TSC1 or TSC2 genes. The changes overactivate a cell-growth pathway called mTOR , allowing LAM cells to grow and spread.

In sporadic LAM, the gene changes are usually acquired within the abnormal cells. They are not present throughout the body and are not generally passed to children.

TSC is different. It is caused by an inherited or newly occurring change in the TSC1 or TSC2

gene throughout the body. A person with TSC can pass the gene change to a child.

The hormone estrogen appears to influence LAM. This may help explain why the condition affects almost entirely women and often appears before menopause. Pregnancy and estrogen-containing medicines may be linked to worsening symptoms in some people, but they do not by themselves cause LAM.

The main recognized risk factors are being female and having TSC. Smoking does not cause LAM, although it can add further lung damage.

Diagnosis

A high-resolution CT scan of the chest is central to diagnosis. It can show the pattern of multiple round, thin-walled cysts spread through both lungs. Other conditions can also cause lung cysts, so the scan must be considered alongside other findings.

A diagnosis may be confirmed when the CT pattern occurs with one or more characteristic features, such as:

VEGF-D is a protein linked to lymph vessel growth. A sufficiently high level can support the diagnosis without a biopsy. A normal or lower result does not completely rule out LAM.

If the diagnosis remains uncertain, a lung biopsy may be considered. This removes a small tissue sample for examination. Because biopsy has risks, specialists generally use the least invasive method that can provide a reliable answer.

Lung function tests do not confirm LAM, but they show how well air moves through the lungs and how effectively oxygen crosses into the blood. These tests are repeated over time to look for change.

Assessment may also include abdominal imaging for kidney angiomyolipomas and lymphatic growths. If TSC is possible, evaluation may look for its other features, such as characteristic skin changes, kidney growths, or changes in the brain.

- Tuberous sclerosis complex
- A kidney angiomyolipoma
- A chylous effusion
- A lymphatic growth called a lymphangioleiomyoma
- A raised blood level of vascular endothelial growth factor D, or VEGF-D
- LAM cells identified in tissue

Treatment and management

Treatment is based on symptoms, lung function, complications, and whether the condition is progressing. People without symptoms or significant lung impairment may initially need monitoring rather than medicine.

Sirolimus is the main medicine used to control LAM. It blocks the overactive mTOR growth pathway. It can slow the loss of lung function and may shrink angiomyolipomas or lymphatic growths. It may also help control chylous fluid collections.

Sirolimus does not remove existing lung cysts or cure LAM. Blood tests and clinical follow-up are needed because it can cause mouth sores, raised cholesterol, swelling, reduced blood cell counts, and a higher risk of infection. Pregnancy planning is important because sirolimus can harm a developing pregnancy.

Other management may include:

A pneumothorax is treated by removing air from around the lung when necessary. Because recurrence is common in LAM, a procedure that helps the lung stick to the chest wall, called pleurodesis , is often considered after the first episode. Previous pleurodesis does not automatically prevent future lung transplantation.

Chylous effusions may be managed with sirolimus, drainage, dietary changes under specialist guidance, or a procedure to seal or redirect leaking lymph vessels.

Kidney angiomyolipomas are monitored with imaging. Growths at higher risk of bleeding may be treated with sirolimus or selective embolization, which blocks their blood supply. Surgery is sometimes needed, but preserving healthy kidney tissue is an important goal.

Lung transplantation may be considered for advanced LAM when breathing is severely limited despite other treatment.

Hormonal treatments such as progesterone or removal of the ovaries are not recommended as LAM treatment. Doxycycline has not been shown to provide reliable benefit.

- Inhaled bronchodilators when airway narrowing improves with them
- Supplemental oxygen when blood oxygen is low
- Pulmonary rehabilitation to improve fitness and breathlessness management
- Vaccination and prompt assessment of respiratory infections
- Support to stop smoking or avoid second-hand smoke

Complications

The most common major complication is pneumothorax. Once it has happened, another episode is more likely.

Other possible complications include:

Complications are not inevitable. Regular monitoring helps identify changes before they cause severe limitations.

- Progressive loss of lung function
- Low blood oxygen and respiratory failure
- Chylous fluid around the lungs or in the abdomen
- Bleeding from a kidney angiomyolipoma
- Reduced bone strength, sometimes worsened by limited activity
- Treatment-related infection or other sirolimus side effects

Prevention

LAM itself cannot currently be prevented. Sporadic LAM results from acquired gene changes, while TSC-LAM is linked to a genetic condition present from birth.

Avoiding smoking cannot prevent LAM, but it helps protect the lung function you have. Avoiding unnecessary estrogen exposure may also be advised because estrogen can influence LAM cells. Do not stop contraception or hormone treatment without discussing safe alternatives with the clinician managing your care.

If you have TSC, recommended lung screening can find LAM before symptoms become noticeable. Screening does not prevent the condition, but earlier detection can guide monitoring and treatment.

Living with it and what to expect

LAM is usually a long-term condition. Its pace differs greatly. Some people have stable lung function for years, while others experience a gradual decline or repeated complications. A single CT scan cannot predict your individual course.

Follow-up commonly includes lung function tests, symptom review, oxygen measurement, and imaging when needed. Kidney angiomyolipomas and other findings may require separate monitoring. The timing of visits is based on your lung function, treatment, and recent rate of

change.

Sirolimus can stabilize lung function for many people while it is being taken, but it does not rebuild damaged lung tissue. If it is stopped, its benefits may not continue.

Regular activity is usually encouraged within your limits. Pulmonary rehabilitation can provide supervised exercise and techniques for managing breathlessness. Some people continue working, travelling, exercising, and caring for families with few adjustments. Others need oxygen, changes in activity, or support with daily tasks as lung function declines.

Air travel is often possible when LAM is stable, but pressure changes may increase concern about pneumothorax. Do not fly with a current or incompletely healed collapsed lung. Discuss travel plans if you recently had a pneumothorax, use oxygen, or have new chest symptoms.

Pregnancy is possible, but it may increase the chance of pneumothorax, chylous effusion, or declining lung function. Pregnancy also affects decisions about sirolimus. Pre-pregnancy advice from clinicians familiar with LAM and high-risk pregnancy can help you understand your personal risks and options.

When to get medical help

Call emergency services for:

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

Seek prompt medical advice for a noticeable but less severe increase in breathlessness, new chest discomfort, blood in the urine, abdominal swelling, or symptoms of infection while taking sirolimus.

Routine follow-up is appropriate for gradual changes in exercise ability, persistent cough, medicine side effects, contraception questions, pregnancy planning, or travel advice. Report changes rather than waiting for the next planned visit if they are affecting daily life.

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

Common questions

Is LAM a type of cancer?

LAM is classified as a low-grade neoplasm, meaning its abnormal cells can grow and spread. However, it does not behave like most cancers, and standard chemotherapy is not used. Treatment targets the mTOR growth pathway.

Is pulmonary LAM inherited?

Sporadic LAM is not usually inherited. TSC-LAM occurs as part of tuberous sclerosis complex, which can be passed from parent to child. Genetic counselling may help if you have TSC or signs that suggest it.

How quickly does LAM get worse?

There is no single timeline. Lung function may remain stable for long periods or decline gradually. Repeated lung function tests give more useful information than one test alone. Sirolimus can slow decline in people who meet treatment criteria.

Can the lung cysts go away?

Existing lung cysts generally do not disappear. Treatment aims to preserve lung function, control abnormal cell growth, and manage complications. A lung transplant replaces severely damaged lungs when the condition is advanced.

Can I fly if I have LAM?

Many people with stable LAM can fly. Flying is not safe with an untreated or incompletely healed pneumothorax. Ask for individual advice after a recent collapsed lung, if you need oxygen, or if you develop chest pain before travel.

Can I have a baby if I have LAM?

Pregnancy is possible, but it can worsen LAM or increase the risk of pneumothorax and chylous effusion. The risk is not the same for everyone. Review lung function, medicines, contraception, and pregnancy plans with a LAM specialist and an experienced pregnancy team.

Sources

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