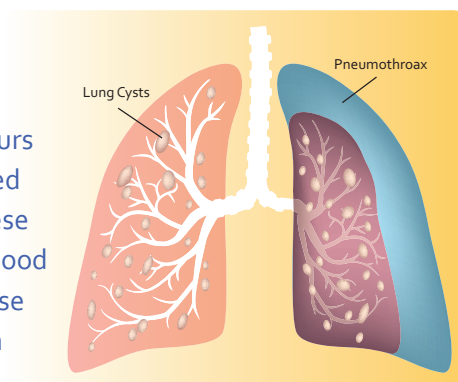


Lymphangiomyomatosis (LAM)

What is LAM?

Lymphangiomyomatosis (lim-FAN-jee-oh-ly-oh-my-oh-ma-TOE-sis), also known as LAM, is a rare lung disease that mainly affects women, usually during their childbearing years. LAM occurs in 3-8 women per million in the general population. LAM is caused by mutations in the tuberous sclerosis complex (TSC) genes. These mutations lead to growth of abnormal cells that spread by the blood stream and make their way into the lungs. Once in the lungs, these cells create holes in the lung tissue (called cysts) that can weaken breathing and the ability to take up oxygen.



What are the symptoms of LAM?

Symptoms of LAM are similar to other lung diseases. Some times patients can be misdiagnosed with asthma, bronchitis, or emphysema. These symptoms include:

- Shortness of breath
- Fatigue
- Lung collapse, also known as a pneumothorax
- Chest pain
- Cough
- Coughing up small amounts blood

How is LAM diagnosed and monitored?

There are many tests that a doctor can do to help make a diagnosis of LAM and to monitor changes, which include:

Imaging

A **chest X-ray** is usually not helpful in diagnosing LAM, as the lung cysts, which are hallmarks of LAM, are often not visible on the chest X-ray. However, chest X-rays are useful in finding a collection of fluid around the lung (pleural effusion) or a collapsed lung (pneumothorax), which are common complications in patients with LAM.

Computed tomography (CT) scan of the chest shows a better image of the lungs and lung cysts. CT of the chest is the most useful test to help make the diagnosis of LAM. One-third of patients with LAM can also have a benign tumor of the kidney, called angiomyolipoma (an-jee-oh-my-oh-ly-PO-ma). This can be picked up by a CT scan of your abdomen.

Blood Testing

About 70% of the patients have an elevated level of a

blood protein called **vascular endothelial growth factor-D** (VEGF-D). Elevated VEGF-D levels can help confirm the diagnosis of LAM without needing a lung biopsy. Blood levels of VEGF-D should be checked before doing invasive procedures, such as lung biopsy.

Lung Biopsy

In most (>70%) patients, the diagnosis of LAM can be confirmed based on clinical and/or laboratory findings and does not require a biopsy. However, a lung biopsy may be needed to confirm the diagnosis of LAM in some cases.

There are two ways to do a lung biopsy to diagnose LAM:

1. **Transbronchial lung biopsy:** A long, flexible tube called a bronchoscope is passed through your nose or mouth into the windpipe and the lungs. Small amounts of lung tissue can be removed using tiny forceps at the end of the bronchoscope and studied for abnormalities. This procedure is commonly done as an outpatient with sedation and local anesthesia. This is a less invasive way to obtain a lung biopsy but may not obtain enough tissue to make an accurate diagnosis. Transbronchial lung biopsy can yield a diagnosis of LAM in 50-60% of patients.
2. **Video-assisted thorascopic lung biopsy** is a more invasive procedure, but provides more lung tissue and is diagnostic in almost all cases. You will be given general anesthesia and have small incisions on the side of your chest. A doctor will pass a thin scope with a camera through the skin and into the chest cavity. While watching on a video screen, biopsies will be taken from the lung tissue for further testing.

Lung Function Testing

The most common way of monitoring LAM is by performing

pulmonary function tests (PFTs). PFTs measure your lungs' ability to breathe and move oxygen by breathing into a machine called a spirometer. These tests are done to assess your lung impairment from LAM and to gauge the rate of disease progression over time. Treatment decisions for starting drug therapy are often based on the results from the PFTs.

How is LAM treated?

There is no cure for LAM at this time. Treatment of LAM centers around a drug called sirolimus (sih-RO-lim-us). Sirolimus may be considered if you have abnormal or worsening lung function, large angiomyolipoma(s), repeated lung collapse, or symptoms from a milky fluid buildup around the lungs. Sirolimus has been shown to stabilize lung function and improve quality of life in people with LAM. Sirolimus is approved by the United States Food and Drug Administration (FDA) for the treatment of LAM. Most people tolerate sirolimus very well. Common side effects of sirolimus include diarrhea, nausea, mouth ulcers, high cholesterol, and leg swelling.

As the disease progresses, some people may require supplemental oxygen. Lung transplantation is an option for some people with advanced disease who do not respond to treatment with sirolimus.

What are the common LAM-related complications?

LAM causes multiple air-filled holes, called cysts, in the lungs. Often these cysts can rupture and cause air to leak outside of the lung, leading to lung collapse. This lung collapse is also known as a pneumothorax. More than half of the patients with LAM experience at least one pneumothorax in their lifetime. Treatment of a pneumothorax may require placing a small tube into the chest cavity to drain the air and re-inflate the lung. Patients with LAM are at a high risk for having multiple episodes of lung collapse and may benefit from procedures aimed at reducing the risk for future events, such as pleurodesis. Also, LAM cells may lead to blockages of the lymph system in the chest and create a collection of milky fluid around the lung, also known as a chylous pleural effusion.

How will LAM affect my lifestyle?

LAM progresses at different rates for each person. In the early stages of LAM, most people can live normally and do not have many symptoms. As the disease progresses, you may find it more difficult to carry out daily activities. Supplemental oxygen may be needed in advanced disease.

You should follow the same healthy diet as the general population. Routine exercise as tolerated is good for your health, and the presence of LAM does not prevent you from doing your routine exercise. It is important to stay up to date on immunizations such as the influenza and pneumococcal pneumonia vaccines. If you are taking sirolimus, avoid live virus vaccines such as the shingles vaccine. Estrogen products, such as contraceptives used for birth control, should also be

avoided. There is a possibility that pregnancy may lead to progression of LAM, so consult your doctor if you are pregnant or considering pregnancy. Do not smoke and avoid exposure to tobacco smoke. Air travel is safe for most patients with LAM. Ask your healthcare provider if you need to take any special precautions.

LAM research programs

Several new treatments for LAM are currently being studied. Patients with LAM may be eligible to join these clinical trials. You should also consider enrolling in a LAM patient registry to help better understand the disease course. More information about these trials and registry can be found at www.clinicaltrials.gov and The LAM Foundation at www.thelamfoundation.org.

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Rx What to Do...

- ✓ Let your healthcare provider know if you are having problems with exercise, shortness of breath, or you cough up blood.
- ✓ Do not smoke and avoid all tobacco smoke exposure.
- ✓ Have your lung function checked regularly.
- ✓ Stay up to date on immunizations, such as influenza and pneumococcal vaccinations.
- ✓ Talk with your doctor to determine if you would benefit from treatment with sirolimus.
- ✓ Develop a pneumothorax emergency management plan with your healthcare provider.
- ✓ Let your healthcare provider know if you are pregnant or are considering getting pregnant.

Healthcare Provider's Contact Number:

Additional Lung Health Information

American Thoracic Society

www.thoracic.org

Fact Sheets on: Oxygen Therapy, Transplantation, Chest Tube Thoracotomy, Bronchoscopy, Pulmonary Function Testing, Tobacco.

ATS Public Advisory Roundtable

<https://www.thoracic.org/patients/par/>

The LAM Foundation

<https://www.thelamfoundation.org>

National Heart Lung & Blood Institute

<https://www.nhlbi.nih.gov/health-topics/lam>

American Lung Association

<http://www.lung.org/lung-health-and-diseases/lung-disease-lookup/lymphangioleiomyomatosis-lam/>

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