Learn About Lymphangioleiomyomatosis

Lymphangioleiomyomatosis (LAM) is a disease that can affect your lungs, kidneys and lymphatic system. The lymphatic system is a circuit that runs through the body carrying cells that maintain your immunity and regulate the nutrients in your blood.

The main finding associated with LAM is the presence of “cysts” in your lung. A cyst is a round space, or “hole” with a thin border that is surrounded by normal tissue.

Key Facts

- LAM affects women of child bearing age, usually in their 30’s and 40’s.
- There are no behavioral conditions or risk factors that are known to cause LAM.
- Though LAM can be aggravated by female hormones and pregnancy, this is not considered a risk factor for the development of LAM.

What is LAM?

The hallmark of LAM is the thickening of the areas surrounding the blood vessels, breathing tubes and lymph channels as well as the lining of the lung. This is the result of an abnormal growth of smooth muscle cells.

LAM is not contagious. When associated with Tuberous sclerosis (TSC), LAM is an inherited disorder that happens when one of the parents pass on an abnormal gene to their children. When LAM presents alone, it is related to the alterations of certain genes that are an important part of cell growth. In rare cases these alterations can happen at random (in approximately 3-7 out of 1 million women).

How LAM Affects Your Body

LAM primarily affects your lungs and your breathing. Occasionally it is associated with another disorder called tuberous sclerosis complex (TSC). When LAM is a part of the TSC, it is more common to have kidney tumors called angiomyolipomas and involvement of the brain that require monitoring and may cause seizure.

How Serious is LAM

LAM can shorten your lifespan, though our experience over the last few decades has shown that patients are living longer than ever before. It is possible that LAM can make you lose your lung function to the point of needing a lung transplant.

The 10-year survival of people living with this disease is 80-90% and a median survival from onset of symptoms is about 30 years.
LAM Symptoms, Causes and Risk Factors
You may be experiencing a variety of symptoms that may be caused by LAM. LAM can affect multiple organs and can have different presentations, so it is sometimes difficult to diagnose.

What are the Symptoms of LAM?
The diagnosis of LAM is strongly suspected when a young female presents with any of the below symptoms. LAM may have a wide array of symptoms including:
- Shortness of breath, especially with exercise
- Cough
- Bloody phlegm

The most common presentation is a pneumothorax, where the lung “pops” and air accumulates around the lung causing it to collapse. In patients with LAM, this occurs due to the bursting of the cysts which are formed in the lungs. Pneumothorax causes sharp chest pains and difficulty with breathing. LAM can also be associated with accumulation of a milky fluid around the lungs called chyle.

What Causes LAM
LAM is caused by abnormal growth of the smooth muscle cells in the lungs. We don't understand well what the triggers for this process are though hormone therapy can aggravate the process.

What are Risk Factors?
There are no known risk factors for LAM and often it is a result of a random genetic mutation or in association with an inherited disorder such as TSC.

When to See Your Doctor
After the diagnosis is made your doctor will arrange for follow up visits to monitor certain things as mentioned in the diagnostic tests below.

However, you should see your doctor any time you have a new, unfamiliar or worsening symptom.

With LAM, as you know you are at risk of tumors called angiomyolipomas in your abdominal organs, most commonly seen in the kidney. As these tumors grow in size they are at risk of internal bleeding that could be fatal and cause symptoms like abdominal pain and dizziness.

Seek prompt medical attention if you experience worsening of your baseline breathing. This could be due to:
- a) Episode of lung collapse (pneumothorax) which can recur
- b) Fluid collection around the lung (chylothorax)
- c) Obstruction of the lymphatic system which can cause fluid overload inside of the lung

Your doctor can then decide whether your symptoms are related to LAM and whether you can be treated at home or if you require hospitalization.
Diagnosing and Treating LAM

What to Expect
Once you are diagnosed with LAM the disease process continues to progress. Suppressive therapy is available but there is no cure and eventually over time the lungs will be replaced by cysts (holes) secondary to muscle infiltration of the lung tissue, airways and the blood vessels in the lungs.

This will lead to progressive respiratory/lung failure at which time lung transplant is the only option available. Expect a close working relationship with frequent follow-ups with your primary care doctor and lung specialist. Various other specialist may be involved at varied stages of this disease.

In the early stages of LAM, you can usually do your normal daily activities. These may include attending school, going to work and doing common physical activities such as walking up stairs. In the later stages of LAM, you may find it harder to be active. Once you are diagnosed with LAM, there should be discussions with your health care provider regarding life planning, pregnancy and birth control.

How LAM is Diagnosed
X-ray is not sufficient to diagnose LAM, so your doctor will order CT scan of the lungs which will identify characteristic cysts. Abdominal ultrasound may reveal tumors in kidney, liver or spleen.

Laboratory tests maybe ordered for levels of vascular endothelial growth factor-D (VEGF-D) which may be elevated in LAM. Your doctor may also order breathing tests to assess lung function. Open-lung biopsy (surgical procedure done in the operating room by a surgeon) or bronchoscopy (a flexible scope with a video camera introduced into your airways through your nose or mouth to inspect your lungs, done by your pulmonologist) may be required to look at lung tissue/cysts.

How LAM is Treated
Currently, no treatment is available to stop the growth of the cysts that occur in LAM. Most treatments for LAM are aimed at easing symptoms and preventing complications. The available treatment options include:

- Oxygen therapy
- Removal of air or fluid from lungs or abdomen to help you breathe better
- Surveillance and treatment of osteoporosis (weak bones)
- Standard vaccinations to prevent respiratory infections
- Bronchodilators (inhalers) to help open airways and improve breathing
- Medications:
  a) Rapamycin (Sirolimus) – Recent studies show that this medication helps to improve lung function, reduce symptoms and may shrink kidney tumors. Sirolimus does have side effects, some of which can be serious. If you have LAM, talk with your doctor about the benefits and risks of this medicine. Note that Sirolimus/Rapamycin therapy is suppressive and thereby might control the disease but is not a cure.
  b) Medroxyprogesterone – no good evidence to support use
  c) Chemotherapy or radiation therapy – no proven role
  d) If your doctor starts you on anti-estrogen therapy be aware that bone and ear health needs to be monitored
- Lung transplantation may be considered in advanced cases as it can improve lung function and quality of life
Living with LAM
LAM is a chronic disease meaning you may have symptoms or require treatment for your lifetime.

Managing LAM
As mentioned above, suppressive therapy is available but once stopped disease process goes back to pretreatment stage.

Oxygen therapy and pulmonary rehabilitation may help to improve your exercise capacity in advanced disease state.

Estrogen containing medications have been shown to worsen LAM and are contraindicated. LAM could be exacerbated by birth control pills and pregnancy causing lung collapse or abortion and preterm labor. Lung transplant is the only curative therapy but recurrence in transplanted lungs is known.

Questions to Ask Your Doctor about LAM
Making notes before your visit, as well as taking along a trusted family member or friend, can help you through the first appointment with your doctor. Here are some questions to ask your health care provider.

1) How do I know that I do not have the genetic condition, the Tuberous sclerosis complex (TSC-LAM)? What tests do we run for the same?
2) At what point and how often will I need imaging studies of my abdomen to assess for tumors in other organs?
3) What are the signs of a pneumothorax/lung collapse?
4) What do I do if my breathing gets worse from what it is?
5) At what point will I need oxygen and will I be a candidate for some type of rehabilitation?
6) When will you start me on treatment for suppression/control of disease?
7) Will I ever be able to conceive? Will my children also be affected if I have TSC-LAM?
8) What form of contraception is safe?
9) At what point should we be discussing about lung transplant and what should I expect from the process?
10) Is LAM known to recur after transplantation?
11) What course do I expect after transplant?
12) Is air travel safe for me?

Finding Support
By now you would know that, as a patient with LAM, you are dealing with a cancer-like (although not cancer) disease. Below are links for for patient support groups.

thelamfoundation.org
lamaction.org