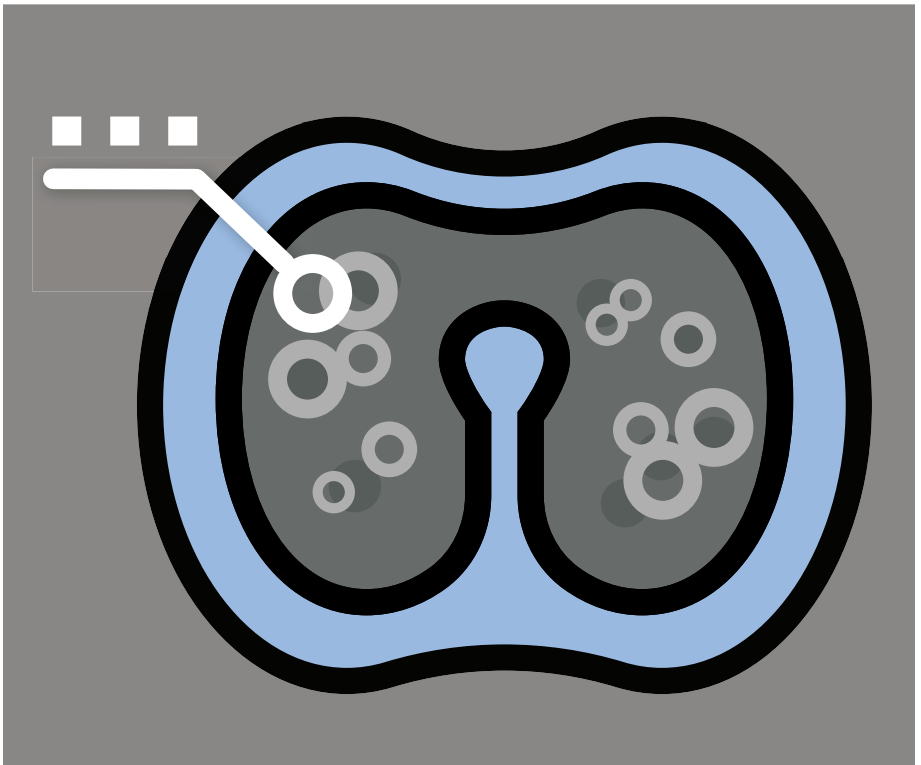




Montreal Chest Institute

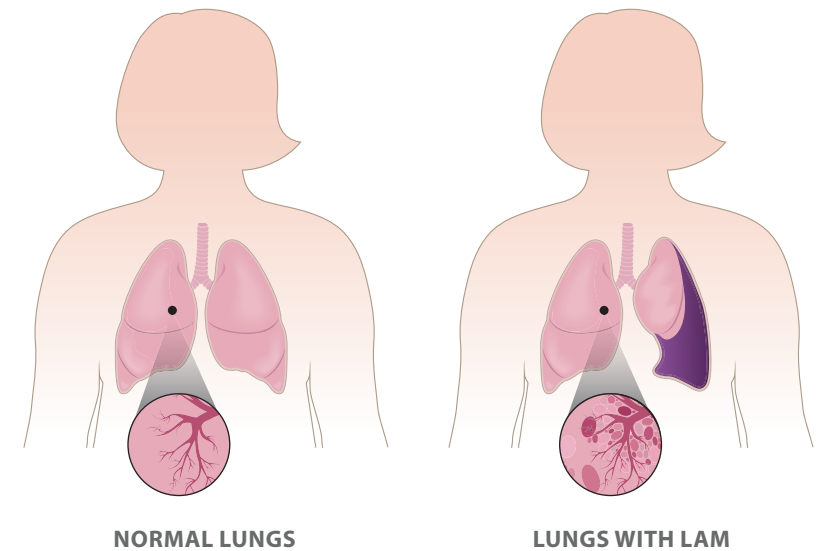
*A Rare Lung Disease //*



A Short Guide to  
***Lymphangiomyomatosis***  
***(LAM)***

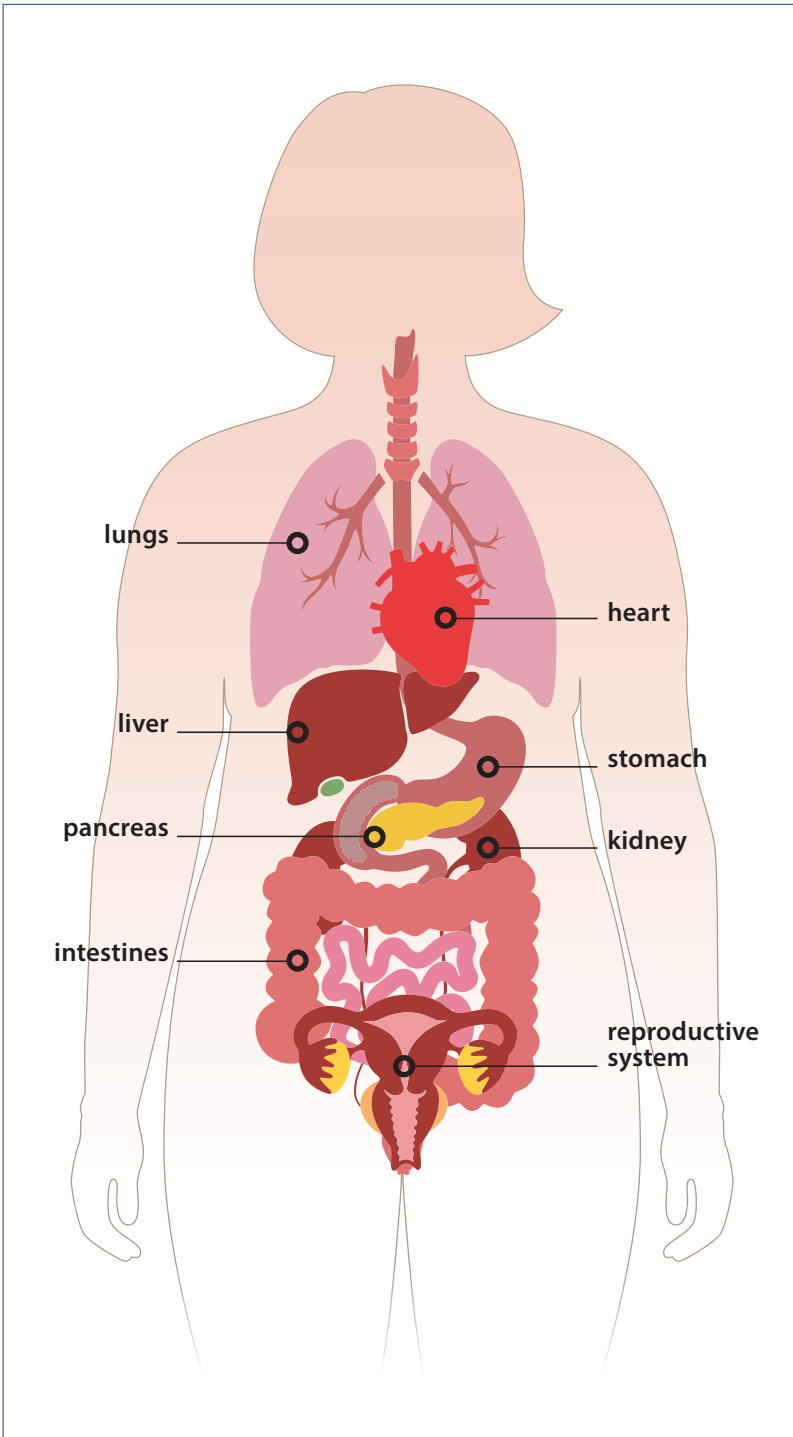
## // What is LAM?

Lymphangioliomyomatosis, or LAM, is a rare lung disease that mainly affects women. Less than 100 women in Quebec are currently diagnosed with LAM. LAM happens most often to women who also have a genetic condition called Tuberous Sclerosis Complex, or TSC. Sometimes, people without TSC can also get LAM. We call this condition “sporadic LAM,” which means that it isn’t caused by another disease the person already has.



LAM affects your lungs and can also affect the kidneys and lymphatic system. The lymphatic system includes the parts of the body that “produce, store, and carry white blood cells that fight infections and other diseases.”<sup>1</sup>

<sup>1</sup> Lymphatic System Definition. National Cancer Institute. [www.cancer.gov/publications/dictionaries/cancer-terms/def/lymphatic-system](http://www.cancer.gov/publications/dictionaries/cancer-terms/def/lymphatic-system)



It is very common for people with LAM to have lung cysts. A lung cyst is a round air-filled sac that has normal lung tissue around it. About one in three people whose lungs are affected by LAM will also have tumours on their kidneys called angiomyolipomas.

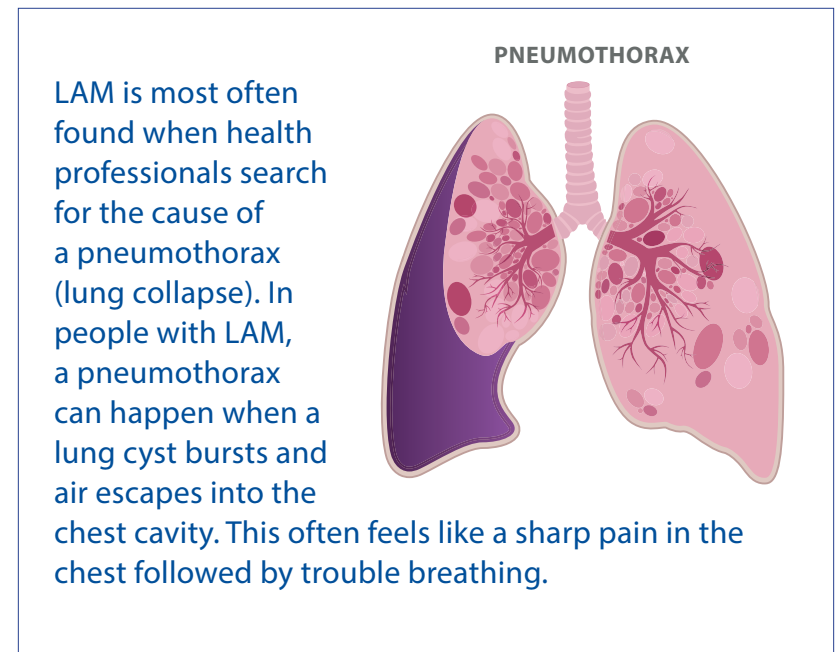
These tumours are usually benign (not cancerous), but they can grow, cause health problems and sometimes become cancerous (malignant), so it is important that your medical team checks on them often.

## // **What are the symptoms of LAM?**

If you have LAM, you may not have symptoms. If you do have symptoms, they may be like the symptoms of many other chronic lung diseases. For this reason, LAM is sometimes misdiagnosed at first as asthma, emphysema or COPD (chronic obstructive pulmonary disease). Emphysema and COPD tend to affect smokers more.

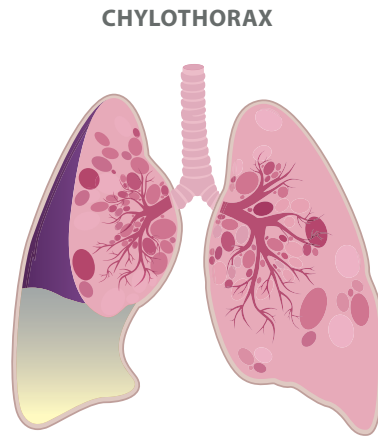
The most common symptoms of LAM are:

- Shortness of breath (trouble breathing)
- Fatigue
- Poor tolerance to effort
- Sudden and severe chest pain from a lung collapse, also called a pneumothorax



Over time, some people with LAM can get shortness of breath as normal lung tissue is replaced by lung cysts, which makes the lungs not work as well. How quickly this happens is different from person to person.

In rare cases, when the lymphatic system is involved, LAM will also cause a build-up of milky, fatty fluid in the chest cavity. This is called chylous pleural effusion. Because chylous effusion almost only happens to people with LAM, it can sometimes help health professionals diagnose this disease.

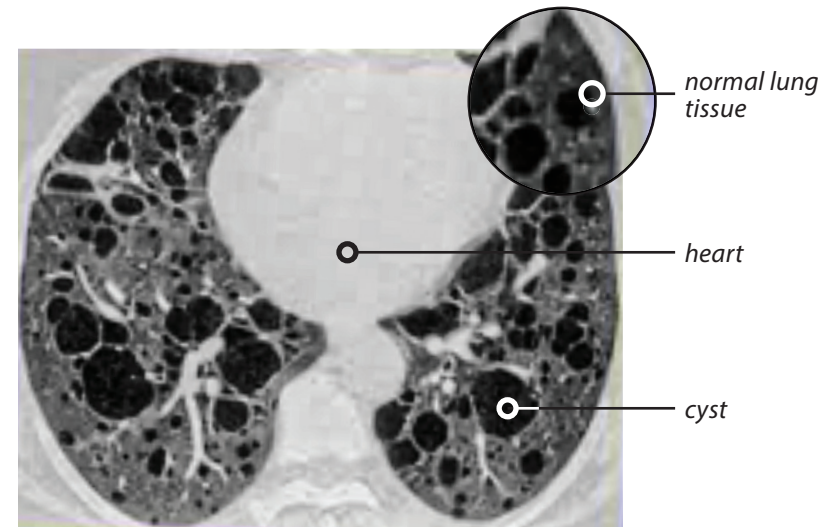


## // What tests are done for LAM?

If you have the symptoms listed in the last section, especially if you are a woman and a non-smoker, health professionals will do tests to check if LAM could be the cause.

### Chest scan

Although a chest x-ray can be helpful to diagnose a lung collapse or a pleural effusion, health professionals will usually ask for a computed tomography (CT) scan of the chest. A chest CT scan can show cysts in the lungs.



These often cannot be seen on regular chest x-rays. If your CT scan results suggest you might have LAM, your health professional may ask you to do more tests.

### Scan of the abdomen (belly area)

Because the kidneys are also often affected, health professionals may request either a CT scan or an ultrasound of the abdomen.



*renal  
angiomyolipoma*

*normal kidney*

### Blood tests

A special blood test can be done to measure the levels of vascular endothelial growth factor-D (VEGF-d). This growth factor is used as a marker for the diagnosis of LAM and is often higher in people with LAM, but not in other lung diseases with lung cysts. If the blood test shows a higher level of VEGF-d, then LAM can be diagnosed without doing a lung biopsy.

### Lung biopsy

A biopsy is when we take a small sample of tissue to check for the presence of a disease. Most LAM patients do not need a lung biopsy.

LAM can usually be diagnosed using other tests. If a lung biopsy is needed in your case, talk to your lung doctor to learn what type of biopsy or procedure is needed and why it is important to do.

### Lung function tests

These tests are done to:

- Check your lung capacity (the amount of air that can move in and out of the lungs when you breathe).
- See what impact the lung cysts might be having on your breathing and health.
- See if the disease is becoming more severe. Changes in lung function are the main way to check the progression of LAM.
- See if your treatment is working.

### If you are diagnosed with LAM

To keep track of your health, you will have regular lung function tests and radiological studies of your lungs and abdomen. These radiological tests may be x-rays, CT scans, MRI scans or ultrasounds.

## // How is LAM treated?

After you are diagnosed with LAM, you will have regular appointments with your lung doctor. You may also see other specialists, such as a urologist, if you have kidney tumours (angiomyolipomas) or a neurologist if you are suspected to also have tuberous sclerosis.

There is currently no cure for LAM. Most treatments for LAM aim to improve your symptoms and help you avoid other health issues.

Some of these treatments include:

- Oxygen therapy for severe disease
- Drains to remove liquid or air in the case of a pleural effusion or a pneumothorax
- Bronchodilators (inhalers), if needed, to help open the airways and improve breathing
- A medication called rapamycin (sirolimus)
- A lung transplant may also be a treatment option for some women with advanced LAM who do not respond well to rapamycin

### Spotlight on rapamycin

Rapamycin is a medication that is sometimes used if:

- Your lung function is getting worse or if it is already very poor
- You have large angiomyolipomas (kidney tumours)
- You have had repeated lung collapses
- You have had symptoms because of a large chylous pleural effusion (build-up of milky, fatty fluid in the chest cavity)

This medication is used to slow the growth of LAM cysts and tumours and to slow down how fast the disease progresses.

Rapamycin is covered by the RAMQ and most private insurance companies. Most people respond well to rapamycin, but side effects are possible. Common side effects include diarrhea, nausea, mouth ulcers, high cholesterol, and leg swelling.

If you are prescribed this medication, your health professional will also ask you to have regular blood tests to check the amount of rapamycin in your system (drug level) and to check for early signs of rarer side effects.

There are also some steps you can take to help improve your symptoms or prevent having more health issues:

- Get vaccinations to prevent common lung infections (such as the flu, pneumonia or COVID-19)
- Avoid hormone therapies and oral medications with estrogen, like the birth control pill. These hormones may play a role in how LAM develops. While it is usually okay to continue using IUDs (intrauterine devices) and creams with estrogen, make sure to talk with your health professional about it first.
- Stop smoking
- Stay at a healthy weight
- Be physically active

## // *How does LAM progress?*

LAM progresses at a different pace for each person. Some people will be diagnosed with LAM by chance and have no symptoms at the time. Other people will be diagnosed with LAM after a lung collapse or having a large pleural effusion.

Most people can go about their day-to-day lives with no limitations, but some people are impacted more severely.

- In the early stages or in mild cases, most people live normally and do not have many symptoms.

When you are diagnosed with LAM, talk to your health professional about life planning, pregnancy, and using hormone therapies or birth control.

- As the disease advances, you may find it more difficult to carry out daily activities. You may need extra oxygen to breathe better.

**// For more information**

There are many trustworthy resources and support groups available to help you. Visit the sites below to learn more about your condition, treatment options, advocacy, and opportunities to take part in research on LAM:

- The LAM Foundation (United States)  
[www.thelamfoundation.org](http://www.thelamfoundation.org)
- LAM Action (United Kingdom)  
[www.lamaction.org](http://www.lamaction.org)
- Regroupement québécois des maladies orphelines  
[www.rqmo.org](http://www.rqmo.org)
- American Thoracic Society  
[www.thoracic.org](http://www.thoracic.org)

**// Notes**

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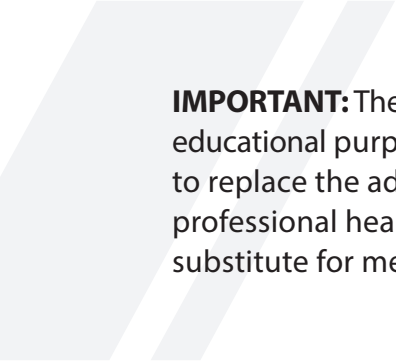


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**IMPORTANT:** The information provided is for educational purposes only. It is not intended to replace the advice or instruction of a professional healthcare practitioner or to substitute for medical care.

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