



What you need to know about...

Lymphangiomyomatosis (LAM)

Lymphangiomyomatosis (LAM) is a rare disease that affects mostly women. LAM causes abnormal cells to grow out of control in some areas of the body, particularly the lungs, kidneys and lymph nodes. This growth creates cysts, which are large clusters of cells. In the lungs, these cysts destroy healthy tissue and make it hard for the lungs to provide oxygen to the body.

Experts do not know the exact number of people who are living with LAM. Although 1,500 cases have been diagnosed, researchers think that up to 250,000 may be going undiagnosed.

What are the symptoms of LAM?

Signs and symptoms include:

- shortness of breath that may get worse over time
- cough that may produce phlegm
- wheezing
- chest pain
- collapsed lung (pneumothorax)
- excess fluid between the chest wall and lung (pleural effusions)
- enlarged lymph nodes
- swelling in the legs, ankles or feet
- abdominal swelling

What causes LAM?

The exact cause of LAM is not known.

Because LAM occurs almost exclusively in women, some think the hormone estrogen might play a role in LAM, although there hasn't been a direct link established. Women with LAM should talk to their doctors before getting pregnant; pregnancy is thought to speed up LAM's progression.

Recent research has found some similarities between LAM and another rare genetic disease called tuberous sclerosis complex (TSC). The kidney growths that occur in people who have TSC are the same as the kidney tumors (called angiomyolipomas) that can be found in women who have LAM. Some women who have TSC get cysts in their lungs that are the same as cysts that women with LAM. Additionally, a defect in the TSC1 or TSC2 genes causes the TSC disease. People with LAM also have abnormal TSC1 and TSC2 genes. Mutations in these genes have been found to cause LAM.

How is LAM diagnosed?

Women often go undiagnosed for years, and because the symptoms are similar to other lung diseases, women are frequently misdiagnosed with asthma, bronchitis or emphysema. Your health care provider will diagnose LAM based on your signs and symptoms, certain tests that show how well your lungs are working and tests to detect LAM cells. A high resolution CT scan, which creates a detailed X-ray of your lungs, is a useful test to detect LAM.

How is LAM treated?

Currently, there is no cure or treatment that has been proven to be effective for LAM, but there is ongoing research. Studies are underway testing the drug sirolimus (rapamycin), which is thought to shrink kidney tumors in women who have LAM. There are treatments available to help relieve symptoms. These may include: oxygen therapy, medicines to relax airway muscles called bronchodilators, hormone therapy, procedures to remove kidney tumors, and lung transplantation.