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Lung involvement in tuberous sclerosis complex (TSC)

Lymphangiomyomatosis (LAM)

LAM is a lung disease caused by genetic variants (previously called mutations) in the *TSC1* or *TSC2* genes. It is characterized by an abnormal growth of smooth muscle cells, especially in the lungs, lymphatic system and kidneys that primarily occurs in women. Unregulated growth of these cells can lead to loss of lung function, accumulation of lymph- rich fluid in the chest and abdomen and growth of tumors in the kidneys. LAM may lead to cystic lung destruction, causing the individual's lungs to lose their functionality. Some cases of LAM occur sporadically and therefore may not have the associated findings of TSC.

Multifocal micronodular pneumocyte hyperplasia (MMPH)

MMPH consists of an overgrowth of pneumocytes (a specific cell type in the lining of the lungs' air sacs). These overgrowths form small nodules that typically do not produce clinical symptoms, but it is nonetheless important to be aware of them as the nodules may be detected.

Management of Lung Manifestations of TSC

Diagnosis and Monitoring

- Chest x-ray
- Pulmonary function tests
- Serum VEGF-D; arterial blood gas
- CT or High-Resolution CT
- Lung biopsy

Interventions

- Sirolimus (MILES study)
- Everolimus
- Inhalers
- Surgical intervention
- Pulmonary transplant

Health considerations

Adult women with TSC:

- Educational counseling on pulmonary manifestations of TSC
- Clinical assessment of potential signs and symptoms of LAM
- Chest CT in all females 18 years or older
- Avoidance of medications with estrogen
- Avoidance of smoking/exposure to second-hand tobacco smoke including use of electronic cigarettes
- Consideration of serum VEGF-D after age 18
- Annual influenza vaccination

Hope no matter how complex



If LAM is diagnosed:

- Receive additional counseling on symptoms and management of pneumothorax and chylothorax.
- Receive guidance regarding pregnancy, smoking, hormone use and air travel.
- Follow-up CT scanning at intervals determined on a case-by-case basis.
- Surveillance by pulmonary function testing depending on disease severity and progression.
- Undergo screening for hypoxemia at rest, or with sleep/exercise with consideration of supplemental oxygen therapy if indicated.
- Consider serum VEGF-D testing
- Consider sirolimus therapy
- Consider referral for lung transplant evaluation for FEV1<30% or disabling dyspnea/hypoxemia
- Annual influenza vaccination
- Avoidance of doxycycline and medications with estrogen
- Speak with a LAM specialist for personalized medical management.

Research resources

Grants and funding



For current information about TSC Alliance grant opportunities, please visit tscalliance.org/grants.

Conferences and meetings

The 2026 TSC World Conference will be held in July in Aurora, CO with a focus on TSC and related disorders including LAM. Clinicians, researchers, and community members from around the world are encouraged to join. Additionally, the TSC Alliance hosts a biennial International TSC Research Conference for medical providers and research scientists. The next one will occur in Fall 2027.

For more information about the TSC World Conference, please visit tscalliance.org/worldconference.

