



# Adults with TSC Newsletter



TSCNow



TSCMatters

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## LAM in Tuberous Sclerosis Complex (TSC)

“LAM in Tuberous Sclerosis Complex (TSC)” was the topic for the Tuberous Sclerosis Alliance’s most recent Adult Topic Call, presented by guest speaker, Karen Agricola, an advanced practice registered nurse and nurse practitioner at the Tuberous Sclerosis Clinic at Cincinnati Children’s Hospital. She helps coordinate initial and follow-up visits, sees patients for follow-up visits and works closely with the doctors and specialists (pulmonary, nephrology, psychiatry, radiology, cardiology, genetics, social work and plastic surgery) to modify treatment plans and help patients navigate their care.

Lymphangiomyomatosis (LAM) is a rare lung disease that affects more women than men. It is characterized by abnormal growth of smooth muscle in the lungs, lymphatic system and

kidneys. There are two forms of LAM: sporadic LAM and LAM in TSC.

“TSC-LAM is the name of one of the pulmonary manifestations that can occur in TSC. Symptoms may be severe regardless if it is caused by TSC-1 or TSC-2,” Agricola said. “It causes cystic destruction of healthy lung tissue, pneumothorax (collapsed lung) and chylous pleural effusions (fluid in lung). LAM cells originate from outside the lung and somehow migrate to the lung, likely through the lymphatic system. Once LAM cells enter the lungs, they secrete enzymes that break down healthy lung tissue leaving cysts or spheres of empty space. This compromises the lung’s ability to move oxygen from the air for transport to the body’s cells as well as being able to exhale carbon dioxide efficiently. Patients with LAM are

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## Adult Resources

### Upcoming Adult Topic Calls:

Thursday, October 24, 2019  
“Open Forum,” presented by Adult Regional Coordinators, at 8 pm Eastern, 7 pm Central, 6 pm Mountain, 5 pm Pacific.

Thursday, November 14, 2019: “Sleep Issues and TSC” presented by Tanjala T. Gipson, MD, Director, TAND Clinic at LeBonheur Children’s Hospital, 8 pm Eastern, 7 pm Central, 6 pm Mountain, 5 pm Pacific.

To participate simply use the toll-free call-in number: 1-866-613-5223 then enter access code: 4334003#. If you have questions or would like to receive handouts (if available) before each call, contact Dena Hook at [dhook@tsalliance.org](mailto:dhook@tsalliance.org) or (800) 225-6872.

Questions? Contact Dena Hook, Vice President, Support Services, at [dhook@tsalliance.org](mailto:dhook@tsalliance.org).

## SAVE THE DATE! Upcoming TS Alliance Events

### Tuberous Sclerosis Alliance Reception

Friday, December 6, 2019, 7 to 9 pm

Plan now to join us for our reception during the 2019 American Epilepsy Society Annual Meeting in Baltimore, Maryland when we’ll highlight public/private partnerships for TSC Centers of Excellence and provide updates on ground-breaking TSC research.

Location and more details to follow.



There final Regional TSC & LAM Conferences is in Los Angeles, California on Saturday, November 2. To learn more about the Regional TSC & LAM Conference Series and to register [click here](#).



Upcoming Step Forward Walks:  
[Saturday, October 26 – Tampa, FL Walk](#)  
[Sunday, October 27 – Cleveland, OH Walk](#)  
To find a walk near you [click here](#).

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at very high risk of these cysts rupturing causing the lung to collapse (pneumothorax). LAM cells can plug up the thoracic duct causing lymphatic fluid to collect in the lung (chylothorax) although this is more likely to occur in LAM not associated with TSC.”

Agricola said, “Because TSC-LAM can cause such severe disease if allowed to progress; it is important that women with TSC be screened early for it. Beginning at the age of 18, women with TSC should be asked at every office visit if they are having any shortness of breath or difficulty breathing with exertion. They should also have a high-resolution computed tomography (HRCT) scan of the chest to look for cysts in the lungs. If there are no cysts, the HRCT should be repeated every 5-10 years.

Agricola said, “If cysts are seen or pulmonary symptoms are present, a referral to a pulmonologist who specializes in LAM should be made for diagnosis and treatment.” Recent research studies showed that rapamycin’s use in LAM stabilized the rate of decline in some lung volumes, improved one’s ability to move air in and

out of the lungs, and improved some quality of life measures. Rapamycin has been FDA approved to treat LAM and is recommended as the first line treatment for people with LAM who have abnormal or declining lung function. Rapamycin is an mTOR inhibitor that is a disease-targeted therapy for TSC. Afinitor is the other mTOR inhibitor that has been FDA approved to treat SEGA (benign brain tumor), renal angiomyolipoma and intractable epilepsy in TSC. Afinitor could be prescribed as an alternative treatment for LAM but is not specifically FDA approved for LAM.

As with any drug, side effects may occur. Mouth sores are the most common. Cholesterol, blood sugar and other functions must be monitored; interactions with other medicines must be reviewed; and vaccinations must be up to date. Sunscreen is a must as sensitivity to the sun is common, and while scuba diving or parachuting are to be avoided, most other activities are not restricted.

For more information and to find research studies, visit [www.thelamfoundation.org](http://www.thelamfoundation.org).

**For more adult resources including previous issues of the Adults with TSC Newsletter, information on reproductive health and contact information for your Adult Regional Coordinator, check out the Adult page on the TS Alliance website [here](#).**



**TSC Now is the TS Alliance’s podcast series!**

[Check out the last episode!](#)

## Additional Resources

*TSC Talks*, a podcast hosted by Jill Woodworth, explores tuberous sclerosis complex and other related issues. Each week Jill talks to parents/caregivers, adults affected by TSC, and other professionals about their experiences managing TSC, the challenges they’ve faced and where they find hope.

[Listen to the latest episode now](#)



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