LUNG FOUNDATION AUSTRALIA

"When you can't breathe... nothing else matters"™

Lymphangioleiomyomatosis (LAM)

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Description: LAM is a rare lung disease almost exclusively affecting women of child bearing age in which bilateral cystic changes occur in the lungs. Abnormal smooth muscle proliferation occurs, resulting in obstruction of small airways, formation of thin-walled cysts, and pneumothorax. LAM is not restricted to the lung and may occur in extrapulmonary sites, particularly the abdomen and pelvis.

Causes: LAM may occur either sporadically (sLAM), or in association with tuberous sclerosis complex (TSC). Although the cause of LAM has not been fully elucidated, current understanding suggests that LAM is probably due to a somatic mutation in the TSC1 or TSC 2 genes.
Clinical Presentations:

- Pneumothorax
- Breathlessness
- Haemoptysis
- Cough
- Chest pain
- Chylothorax

Exacerbations of LAM have been reported with pregnancy

Investigations:

- High resolution CT scan of the chest shows multiple thin walled cysts. Abdominal cuts may show angiomyolipomas (AMLs) in approximately 50% of women, or abdominal lymphangioleiomyomas in 10-15%.
- Lung function tests show airflow obstruction, gas trapping and a low DLCO. A significant bronchodilator response may also be present.

Treatment:

- There is no level 1 evidence for any treatment in LAM, but doxycycline (an MMP inhibitor) or mTOR inhibitors (e.g. rapamycin) may be of benefit and trials are currently underway
- Symptomatic treatment e.g. pleurodesis, bronchodilators
- Lung transplantation

References:

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