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LYMPHANGIOLEIOMYOMATOSIS: A CASE REPORT

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Objective: INTRODUCTION
Lymphangioleiomyomatosis (LAM) is an orphan lung disease often associated with tuberous sclerosis complex (TSC). TSC is an autosomal dominant genetic disease caused by inactivating mutations in the TSC1 and/or TSC2 genes that stimulate mTOR enzyme. So far treatment option is sirolimus (rapamycine), a mTOR inhibitor which is originally used to prevent organ rejection. Sirolimus may reduce volume of angiomyolipomas and prevent decline of FEV1, but there are still not enough data regarding other TSC and LAM complications, nor for how long the treatment should last. Taking in consideration its toxicity, it is important to evaluate benefit of long-term treatment. So far the recommendation for treatment continuation is rapid decline of FEV1 (rather than pneumothorax).
CASE REPORT
We present a case report of 40 year old female patient with tuberous sclerosis and LAM with the history of bilateral pneumothoraces for which VATS pleurodesis was performed at the age of 31. Patient also has skin angiofibromas, enamel pits and renal angiomyolipomas. Her mother had TSC, LAM, and renal failure caused by rupturing of renal angiomyolipomas.
Diagnosis of TSC and LAM was made at the age of 36 and treatment with sirolimus was initiated.
Patient was treated with sirolimus for two years and since there was no reduction in size of angiomyolipomas and no improvement in lung function, it was decided that the therapy should be ceased for three months and to continue follow up. Three weeks after cessation of sirolimus, the patient developed series of bilateral

pneumothoraces (Picture 1 and 2) and thoracic drainage followed by VATS pleurodesis was performed. Sirolimus was reintroduced and since then the patient did not have pneumothorax and the skin angiofibromas have markedly reduced. Lung function tests showed moderate airflow obstruction and decreased DLco, but have remained stable during long-term sirolimus therapy.

CONCLUSION

Effective treatment for LAM is limited. Available options are mTOR inhibitors and lung transplantation.
In this case report we showed that cessation of sirolimus can trigger recurrent pneumothoraces and consequently decrease of lung functions. Long-term therapy should be considered in such patients.