

# Tuberous Sclerosis Complex with Lung Involvement

Arthur Robert Kurzbuch<sup>1</sup> Jean-Yves Fournier<sup>1</sup>

<sup>1</sup>Department of Service de Neurochirurgie, Hôpital du Valais, Centre Hospitalier du Valais Romand (CHVR), Hôpital de Sion, Sion, Switzerland

**Address for correspondence** Arthur Robert Kurzbuch, MD, Department of Service de Neurochirurgie, Hôpital du Valais, Centre Hospitalier du Valais Romand (CHVR), Hôpital de Sion, Avenue du Grand-Champsec 80, CH-1951 Sion, Switzerland (e-mail: kurzbuch@web.de).

J Neurosci Rural Pract 2020;11:211

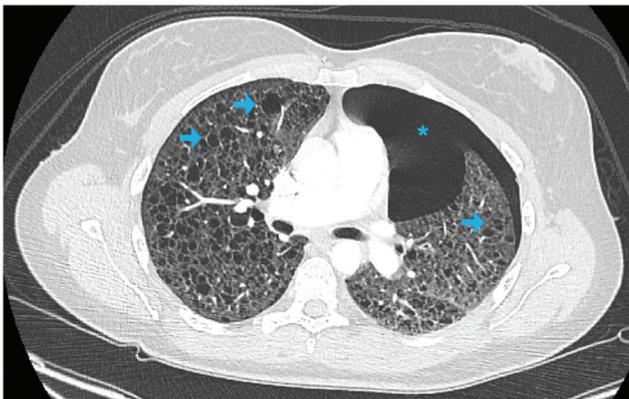
A 31-year-old woman, nonsmoker, known for tuberous sclerosis complex (TSC) underwent uneventful endoscopic resection of a growing subependymal giant cell astrocytoma localized in the left lateral ventricle. Two days after the operation the patient developed dyspnea. Computed tomography of the chest revealed a left-sided pneumothorax (blue asterix) and diffuse cystic changes throughout the lung parenchyma (blue arrows) consistent with pulmonary lymphangiomyomatosis (LAM; ► **Fig. 1**). TSC is an autosomal dominant disorder, resulting from mutations in chromosome 9 (TSC1 encoding hamartin) or chromosome 16 (TSC2 encoding tuberin), that manifests with multisystem involvement: skin, brain, heart, kidneys, and lungs. LAM affects predominantly women and is characterized by proliferation of abnormal smooth-muscle cells in the lung parenchyma that are responsible for cystic changes. Clinically, patients present with dyspnea and pneumothorax. The pneumothorax required draining for 4 days and the patient was discharged home 2 days later without further complications.

## Conflict of Interest

None declared.

## References

- 1 Boone PM, Scott RM, Marciniak SJ, Henske EP, Raby BA. The genetics of pneumothorax. *Am J Respir Crit Care Med* 2019;199(11):1344–1357
- 2 Gupta N, Henske EP. Pulmonary manifestations in tuberous sclerosis complex. *Am J Med Genet C Semin Med Genet* 2018;178(3):326–337
- 3 Henske EP, Józwiak S, Kingswood JC, Sampson JR, Thiele EA. Tuberous sclerosis complex. *Nat Rev Dis Primers* 2016;2:16035



**Fig. 1** Axial computed tomography of the chest shows diffuse cystic changes throughout the lung parenchyma consistent with pulmonary lymphangiomyomatosis (blue arrows) and a left-sided pneumothorax (blue asterisks).