Tuberous Sclerosis Complex with Lung Involvement

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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 asterisk) and diffuse cystic changes throughout the lung parenchyma (blue arrows) consistent with pulmonary lymphangioleiomyomatosis (LAM; ► **Fig. 1**). TSC is an autosomal dominant disorder, resulting from mutations in chromosome 9 (TSC1 encoding hamartin) or chromosome 16 (TSC2 encoding tuberlin), 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, Jóźwiak 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 lymphangioleiomyomatosis (blue arrows) and a left-sided pneumothorax (blue asterisks).