Persistent cardiac rhabdomyoma in an adult with tuberous sclerosis

Pierre-Yves Courand1*, Martine Barthelet 2, Jean-François Cordier 3,4, and Vincent Cottin 3,4

1Department of Cardiology, Louis Pradel Hospital, Hospices Civils de Lyon, 69 317 Lyon cedex 04, France; 2Department of Echocardiography, Louis Pradel Hospital, Hospices Civils de Lyon, Lyon, France; 3Department of Respiratory Diseases, National Reference Center for Rare Pulmonary Diseases, Louis Pradel Hospital, Hospices Civils de Lyon, Lyon, France; and 4University Claude Bernard Lyon I, INRA, UMR754 INRA-Vetagrosup EPHE IFR 128, Lyon, France

* Corresponding author. Tel: +33 4 72 35 78 72; fax: +33 4 72 35 70 49, Email: pycourand@hotmail.com

A 28-year-old woman was referred to our hospital for recurrent pneumothorax. Her past medical history was remarkable for tuberous sclerosis diagnosed at the age of 2 years, presenting with epileptic status associated with mental retardation. On admission, she was found to have severe pulmonary lymphangioleiomyomatosis (Panel A) with end-stage multiple cystic lung disease complicated with recurrent pneumothorax requiring chest tube drainage and surgical pleurodesis. Unenhanced cerebral computed tomography showed bilateral calcification along the walls of the lateral ventricles characteristic of cerebral tubers (Panel B). She also had skin and renal involvement including hypopigmented macules, facial angiofibromas (Panel C), multiple liver angiomyolipomas, and a large bilateral renal angiomyolipoma (Panel D). As clinical diagnostic criteria for tuberous sclerosis were met, gene mutation analysis was not performed. Pulmonary involvement led to chronic respiratory failure requiring long-term supplemental nasal oxygen therapy. Transthoracic echocardiography demonstrated two cardiac tumours involving the interventricular septum and the inferior wall of the left ventricle, however not causing left ventricular inflow or outflow tract obstruction (Panel E). In this setting, the diagnosis of cardiac rhabdomyoma was made. During the clinical course, the patient remained free of any cardiac manifestation. She declined lung transplantation and later died of chronic respiratory failure. Autopsy was declined.

Tuberous sclerosis is a rare multisystem disorder with autosomal dominant inheritance (mutations in either one of the two tumour suppressor genes TSC1 and TSC2). The persistence of cardiac rhabdomyomas in adults with tuberous sclerosis is exceedingly rare. Most tumours regressed before birth or disappeared by the age of 4 years.

Funding. Comité National contre les Maladies Respiratoires.

Published on behalf of the European Society of Cardiology. All rights reserved. © The Author 2012. For permissions please email: journals.permissions@oup.com