Cardiac Involvement in Tuberous Sclerosis

Afección cardiaca en la esclerosis tuberosa

Ernesto Valero,* Gema Miñana, and Francisco J. Chorro

Servicio de Cardiología, Hospital Clínico Universitario, Valencia, Spain

Tuberous sclerosis or Bourneville’s disease is a genetic disorder of autosomal dominant inheritance with high frequency and variable expression that increases predisposition for benign tumors in neuro-ectodermal tissues, particularly of the central nervous system and the skin. It is a very rare disease (5 to 7 per 10,000 live births) that is characterized by the triad of: skin lesions, seizures, and mental retardation. Among its extracutaneous manifestations, the cardiac involvement stands out (present in 30%-50% of cases), always in the form of cardiac rhabdomyomas.

We report the case of a 29-year-old woman who was diagnosed of tuberous sclerosis in childhood, with known cerebral, renal, and dermatological effects, referred to our hospital for echocardiography. The physical examination revealed the presence of numerous facial angiofibromas. The electrocardiogram was normal; we observed, in the parasternal view (Figs. 1A and B), two nodular, homogeneous, well defined, hyperechoic masses, located at the interventricular septum that protruded into both ventricles. In the apical 4-chambers view we observed a third mass of similar characteristics but smaller in size (Fig. 2), as well as a right ventricular moderator band that was thickened and hyperechoic if compared with normal features (Fig. 3).

Due to the patient’s clinical context, we concluded that the intracardiac masses found were three intramural rhabdomyomas located at the interventricular septum.

* Corresponding author:
E-mail address: ernestovaleropicher@hotmail.com (E. Valero).
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