Cardiac Rhabdomyomas in Tuberous Sclerosis (Bourneville's Disease)

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This report describes the case of a 14-year-old boy with moderate mental retardation who had had seizures since infancy. At birth it was noticed that he had hypochromic ash leaf-shaped macules in the trunk. Two years later, facial and ungual lesions appeared (Figs 1 and 2). When he was 7 years old, a diagnosis of tuberous sclerosis (Bourneville's disease) was made. At 13 years old, a cranial computed axial tomographic scan and echocardiogram revealed characteristic lesions related to Bourneville's disease (Figs 3 and 4). The patient has never had cardiac symptoms, and physical examination and ECG are normal.

Fig 1. Photograph showing facial angiofibromas involving cheeks, nose, and chin, with relative sparing of the upper lip.

Fig 2. Photograph showing fibromas of nails.

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Fig 3. Cranial computed axial tomographic scan showing calcified subependymal glial nodules in the lateral walls of the lateral ventricles.

Fig 4. Echocardiogram in apical four-chamber view with several medium-size (0.8- to 1-cm) intracardiac masses (probably rhabdomyomas) in the left ventricle located in the apex and interventricular septum. There is also a large mass (3.5×1 cm) attached to the lateral wall of the left ventricle.
Images in cardiovascular medicine. Cardiac rhabdomyomas in tuberous sclerosis (Bourneville's disease).
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