A 2-MONTH-OLD infant, born by spontaneous vaginal delivery, was admitted for treatment of cellulitis of 1 toe as a result of a hair tourniquet. He was 3 kg at birth after a full-term pregnancy. His mother was aged 26 years, gravida 2, para 1, aborta 0 and had a seizure disorder for which she received divalproex sodium daily. She had learning disabilities and required special education. The infant’s father, aged 60 years, is healthy. The infant was observed in the hospital for 1 week for an irregular heartbeat. He was well until he was admitted for treatment of cellulitis. Findings from physical examination revealed cutaneous lesions (Figure 1). He had a single café-au-lait lesion on his left arm. A cerebral magnetic resonance imaging scan (Figure 2 and Figure 3) and an echocardiogram (Figure 4) were obtained. The cellulitis resolved after administration of antibiotic therapy, and he was discharged. At age 6 months, he developed infantile spasms.
Tuberous Sclerosis

Figure 1. Hypopigmented lesions on the skin of the affected infant.

Figure 2 and Figure 3. Magnetic resonance image shows periventricular nodules and a giant cell astrocytoma of anterior horn of right ventricle.

Figure 4. Echocardiogram shows right atrial rhabdomyoma.

Tuberous sclerosis (TS) is a condition that is inherited as an autosomal dominant trait and characterized by hamartomas involving multiple organ systems. The incidence is 1 in 10,000 births. Clinical presentations vary widely. Cutaneous involvement is a frequent sign of TS. Hypomelanotic macules are the earliest skin manifestations (Figure 1 and Figure 2) and are most apparent when the surrounding skin is dark. They can be identified clearly with use of a Wood light. The shape is polygonal or ash-leaf, and on occasion they occur as groups of numerous small (1-3 mm) white macules.1 Other cutaneous lesions include connective tissue hamartomas with facial angiofibroma (adenoma sebaceum) present in at least half of all patients with TS. They are localized over the nasal bridge and cheeks in a butterfly distribution and appear between age 5 years and puberty. Ungual fibromas, Shagreen patch, and forehead fibrous plaque1 are cutaneous findings.

Severe central nervous system involvement is characteristic of TS. Cortical tubers and an astrocytoma are demonstrated in Figure 3 and Figure 4. The brain hamartomas may cause complex or partial seizures that evolve over time.1 The tubers, which consist of sclerotic tissue, are located throughout the cerebral hemispheres. These tubera are histogenetic malformations of both neuronal and glial elements with decreased neurons, increased glia, and abnormal giant heterotopic cells. Approximately 6% to 14% of patients with TS will develop giant cell astrocytomas during the first 2 decades of life.2 Contrast-enhanced magnetic resonance images or computed tomographic (CT) scans help to distinguish a giant cell astrocytoma from other cerebral lesions. Of children with infantile spasms, 10% will have TS.3 Mental retardation, autism, attention-deficit/hyperactivity disorder, or a combination of these conditions are seen in TS.4 Cerebrovascular anomalies are rare, but a few cases of cerebral arterial ectasia and giant fusiform aneurysm formation in children have been reported. Magnetic resonance angiography may be used for diagnosis, which prevents obtaining biopsy specimens of a vascular lesion.2 The cardiovascular system is affected in TS. Multiple clinically silent cardiac rhabdomyomas are found in affected infants (Figure 5). The masses are most often located in the ventricles where they may cause arrhythmias or outflow obstruction of one or both ventricles.1 The rhabdomyomas have been found to resolve spontaneously in infancy or by early adolescence.1 Renal involvement, common in TS, is characterized by renal cysts. The cysts are bilateral, multiple, of varying sizes, and usually asymptomatic.1 Although they mimic autosomal dominant cystic kidney disease, they can be distinguished histopathologically. Solid masses are angiolipomas, which are vascular tumors consisting of smooth muscle, adipose tissue, and fibrous tissue. The angiolipomas are present more of-