A newborn with hypopigmented skin macules, cardiac rhabdomyomas, and cerebral magnetic resonance imaging (MRI) demonstrating cortical tubers and subependymal nodules (Figure, A) was diagnosed with tuberous sclerosis complex (TSC). Clusters of flexor spasms began at 3 months of age, infantile spasms was diagnosed. Treatment was commenced with vigabatrin, an irreversible inhibitor of γ-aminobutyric acid transaminase, and the spasms ceased within 1 week. Routine follow-up MRI demonstrated the interval development of asymptomatic abnormalities, including restricted diffusion in the bilateral brainstem, basal ganglia, and thalami (Figure, B). After 1 year of successful treatment, the vigabatrin was discontinued and follow-up MRI demonstrated full resolution of these abnormalities (Figure, C).

TSC is a genetic disorder characterized by hamartomatous lesions in multiple organ systems. Infantile spasms are common in TSC, occurring in one-third of patients. Vigabatrin is considered first-line therapy for infantile spasms in patients with TSC due to its remarkable effectiveness (>95%). These striking drug-related MRI abnormalities due to vigabatrin therapy occur primarily in infancy, are dose dependent, and are apparently asymptomatic and fully reversible. The pathophysiology is not known, but studies in animals suggest the contribution of intramyelinic edema, and the high risk confined to infancy suggests a developmental role of brain maturation.

Bilateral MRI abnormalities in a similar distribution (brainstem, basal ganglia, thalami) can also be seen in metabolic, ischemic, infectious, and toxic etiologies. The patient history, to include concurrent medications, is especially pertinent in the clinical interpretation of these MRI findings.

References

Figure. A, Axial T2-weighted MRI of the brain at 1 month of age. Multiple hypointense subependymal nodules (white arrow) and areas of cortical thickening and hypointense signal (black arrow) representing calcified tubers, are present. B, Axial diffusion-weighted MRI of the brain at 13 months of age. Abnormal hyperintense signal is present in the bilateral brainstem, basal ganglia, and thalami. C, Axial diffusion-weighted MRI of the brain at 27 months of age. The previous brainstem, basal ganglia, and thalamic abnormalities have resolved.