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Utility of early single photon emission computed tomography (SPECT) in neonatal gelastic epilepsy associated with hypothalamic hamartoma.

[DiFazio MP](#), [Davis RG](#).

Department of Child and Adolescent Neurology, Walter Reed Army Medical Center, Washington, DC, USA.

Gelastic epilepsy, or laughing seizures, is a rare seizure manifestation often associated with hypothalamic hamartoma. This seizure type is well described in older children and adults, but has only rarely been reported in neonates, oftentimes recognized in retrospect when the children are older. We report a child diagnosed at 3 months of age with a large hypothalamic mass after evaluation for spells occurring since birth. The spells were characterized by bursts of hyperpnea, followed by repeated "cooing" respirations, giggling, and smiling. These spells were recognized soon after birth in the delivery room, and occurred at 15-20 minute intervals. They did not interrupt feeding and occurred during sleep. On referral to our center, the patient was noted to be thriving, with normal medical and neurologic examinations except for his spells. The laboratory evaluation was normal, as were endocrine and ophthalmologic evaluations. Neuroimaging was performed, with magnetic resonance imaging demonstrating a large 2.8-cm isodense, nonenhancing hypothalamic mass. Electroencephalogram was abnormal, demonstrating bi-frontal sharp and spike-wave discharges. Video-EEG did not demonstrate ictal discharges associated with the patient's spells. Single photon emission computed tomography (SPECT) demonstrated dramatic ictal uptake in the area of the tumor, with normalization during the interictal phase. Partial excision of hamartomatous tissue has minimally improved the spells. In conclusion, this patient manifested an unusual, early presentation of a rare seizure type. SPECT scanning confirmed the intrinsic epileptogenesis of the hamartoma, further justifying a surgical approach to such patients. Early surgical intervention is probably indicated in an attempt to minimize or prevent the cognitive and behavioral sequelae commonly seen with this seizure type.

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