A 14-month-old boy presented with loss of developmental milestones and tonic spasms following a diarrheal illness. He was born to nonconsanguineous parents and had mild motor delays. Examination was remarkable for macrocephaly, axial hypotonia, and asymmetric dystonic posturing of neck, trunk, and extremities. Brain MRI (figure) and elevated glutaryl carnitine on tandem mass spectroscopy were diagnostic of glutaric aciduria type 1 (GA-1).1

Widening of the sylvian fissures, mesencephalic cistern, and enlarged pretemporal subarachnoid spaces are cardinal MRI features.2 GA-1 should be considered in any infant or young child with acute encephalopathy in the presence of macrocephaly and extrapyramidal manifestations.

AUTHOR CONTRIBUTIONS
Dr. Singh participated in acquisition of data and drafting/revising the manuscript. Dr. Goraya participated in acquisition of data, study concept or design, analysis or interpretation of data, and drafting/revising the manuscript. Dr. Ahluwalia participated in analysis or interpretation of data and study supervision. Dr. Saggar participated in analysis or interpretation of data and study supervision.

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