



## Letter to the Editor

**Right frontal lobe encephalomalacia in an adult propionic acidemia patient with neuropsychiatric manifestations**

**Keywords:**

Propionic acidemia  
Metabolic stroke  
Encephalomalacia  
Frontal lobe  
Neuropsychiatric

Dear Sir,

We describe a 29 year old patient with propionic acidemia presenting with a sudden onset of neuropsychiatric symptoms. The diagnosis of propionic acidemia was made by a fibroblast enzyme assay at the age of 5 weeks during an episode of coma and acidosis. He had several episodes of metabolic decompensation in early childhood leading to global developmental delays. The patient was lost to follow up after 8 years of age. He reestablished care at 26 years of age. No abnormal behavior was noted at that time. He was noted to have a diminished tone of extremities and was reported to use the left upper extremity less frequently. He was living in a group home and taking carnitine supplements. He was reportedly on a low protein diet supervised by a nutritionist at the group home. After reestablishing care, his carnitine supplementation and diet were optimized. Subsequently his clinical examination was unchanged and his laboratory parameters (plasma amino acids, urine organic acids, plasma free and total carnitine, and plasma acylcarnitine profile) suggested good metabolic control. The patient had a sudden onset of abnormal behavior at 28 years and 9 months of age. It consisted of outbursts of aggressive behavior. He was noted to be violent and would hit his caregivers and other residents at the group home. He was mostly confined to himself and lost interest in things he used to like such as gardening. He had frequent spells of crying for no apparent reason. He would wake up early in the morning and enter other residents' rooms and wake them up too. He also developed obsessive behaviors such as playing with water, putting shoes in grocery bags, playing with toilet paper, collecting towels, etc. He would repetitively ask the same question. He appeared anxious at his new baseline. He had poor sleep. His appetite diminished and he had significant weight loss. He was hospitalized and evaluated for metabolic decompensation during an episode of behavioral outburst. His evaluation was unremarkable. He was reported to have staring spells but EEG was not suggestive of seizure. During an outpatient visit at the metabolic clinic, he was found to have a diminished tone and brisk reflexes in all extremities but there were worsening contractures and diminished movement of the left upper and lower extremities which were new findings. Hence, MRI of the brain was obtained which showed right frontal lobe encephalomalacia (Fig. 1).

The reason for the current behavioral abnormality is unclear. There is no evidence of acute or chronic metabolic decompensation. The

possibility of a complex partial seizure was considered but his EEG was not suggestive, neither was there a response to anticonvulsants. It may have been precipitated by the frontal lobe encephalomalacia as right frontal lobe lesions are associated with disorganized behavior and anxiety [1,2]. However, encephalomalacia is most likely a result of previous metabolic stroke and may not explain acute symptoms.

Metabolic stroke in propionic acidemia usually involves basal ganglia and sometimes results in cystic changes [3]. Frontal lobe encephalomalacia has not been reported in patients with propionic acidemia thus far. Also, very few patients with propionic acidemia and psychiatric symptoms are reported [4,5]. As patients with propionic acidemia are growing older we are likely to see more long term issues including neuropsychiatric manifestations. A sudden onset of behavioral changes should lead to investigation for metabolic decompensation and seizures. Neuroimaging should be considered for the possibility of structural brain lesions resulting from previous brain injury which may be either a causative or contributory factor.

**Author contribution**

Dr. Pankaj Prasun was involved in patient care, laboratory interpretation, initial drafting of the manuscript, and revisions of each draft.

Ms. Lauren Bailey was involved in patient care and revisions of each draft.

Dr. Priya Kishnani supervised the case report, was involved in patient care, laboratory interpretation, and revising the manuscript critically for important intellectual content.

All authors have approved the article as it is written. This work was carried out at Duke University Medical Center.

**Declaration of conflicting interests**

Dr. Pankaj Prasun, Ms. Lauren Bailey, and Dr. Priya Kishnani have no potential conflicting or competing interests that could in any way affect the conduct of the study, interpretation of results, or preparation of the manuscript.

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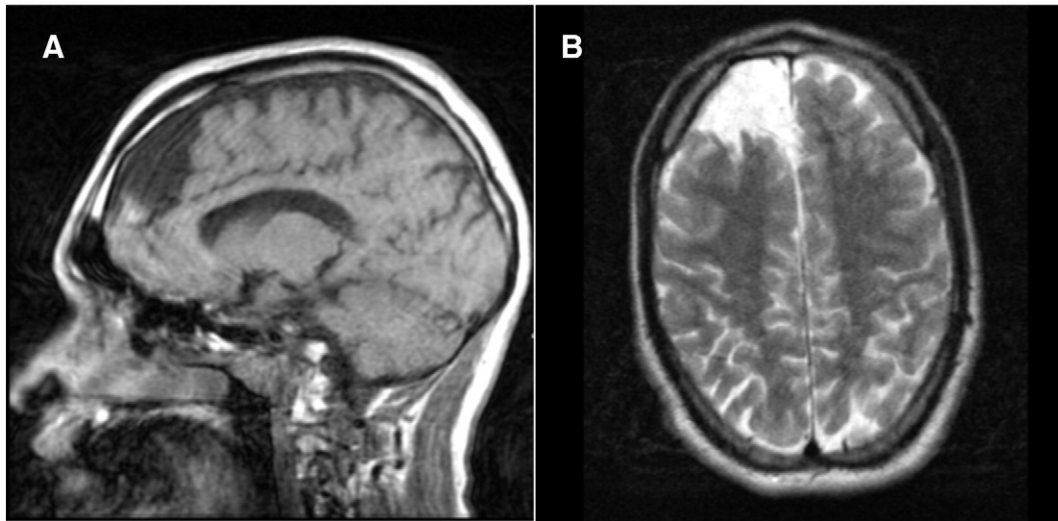
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**Ethical consideration**

This clinical report is a retrospective clinical observation that does not require ethics committee approval at this institution.

There are no prior publications of this manuscript.

Each author listed on the manuscript has seen and approved the submission of this version of the manuscript and takes full responsibility for the manuscript.



**Fig. 1.** Magnetic resonance imaging (MRI) of the brain of a 29 year old patient with propionic acidemia and neuropsychiatric manifestations showing right frontal lobe encephalomalacia in sagittal T 1 (A) and axial T 2 (B) scans.

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