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Dear Editor,

GLUTARIC ACIDURIA TYPE 1 PRESENTING AS SUBDURAL HAEMATOMA

Glutaric aciduria type 1 (GA1) is an autosomal recessive disorder caused by deficiency of glutaryl CoA-dehydrogenase. The metabolism of lysine, hydroxylysine and tryptophan is blocked, leading to accumulation of glutaric acid (GA) and increased urinary concentrations of GA and 3-hydroxy GA. Onset usually involves an acute encephalopathic episode in a macrocephalic infant, associated with fever. Unlike other organic acidemias, metabolic and lactic acidosis, hyperammonemia and hypoglycaemia are rarely present. Subsequently, there is gross motor delay with marked dystonia-dyskinesia. Subdural haematoma is a rare presenting feature of GA1.[1] We report a case who presented with subdural haematoma and diagnosed as GA1.

A 17-month-old girl presented to emergency department with minor head trauma. She had fallen down a chair. Her initial neurological examination was completely normal in the emergency unit. She had a generalised seizure. Computerised brain tomography revealed subdural haematoma and she was operated on. During follow-up, her muscle tone increased gradually and previosuly achieved developmental milestones lost. At the age of 24 months, she was unable to sit and walk. Brain magnetic resonance imaging showed bilateral temporal atrophy (Fig. 1).

In tandem mass spectrometry, low free carnitine [(5.6 µmol/L) (N:10–60)] and high C5 DC glutarile carnitine [(0.71 µmol/L) (N:0.0–0.40)] levels were detected. Urine organic acid analysis revealed a 20-fold increase in the GA level. With the help of these findings, GA1 was diagnosed. The child was commenced on a reduced lysine/tryptophan diet with carnitine supplementation.

GA1 is a relatively rare disease and the 'typical' presentation is within the first 12 months of life of an acute metabolic encephalopathic crisis followed by loss of motor skills and development of a dystonic-dyskinetic movement disorder.[1] The presence of acute and chronic subdural collections has been reported in 20% to 30% of patients with GA1 and might be the presenting feature.[2]

The pathogenesis of subdural haematomas in GA1 is unclear. It was hypothesised as caused by expanded extra-axial cerebrospinal fluid spaces that result in stretching of the bridging cortical veins. Consequently, patients are prone to developing subdural haemorrhages after minor trauma.[3]

In the absence of a history of adequate trauma, the presence of subdural collections of may lead clinicians to suspect non-accidental trauma in a child with undiagnosed GA1.[4] In our case with minor trauma, subdural haematoma was detected by brain tomography and magnetic resonance imaging showed temporal atrophy, which is the most frequent radiological feature of GA1. The diagnosis of GA1 explained the subdural haematoma.

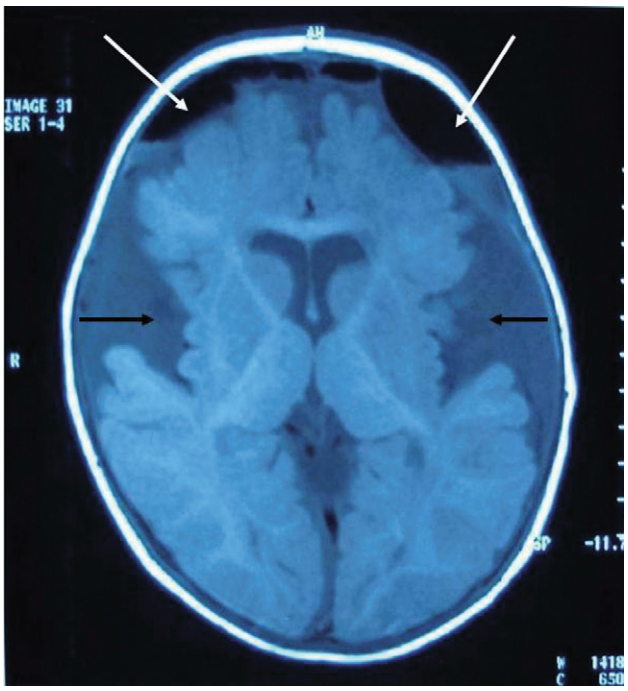


Fig. 1 T1-weighted axial image showing bifrontal subdural (white arrows) haematoma and bitemporal atrophy (black arrows).

The case is being presented to emphasise that subdural haematoma might be an initial feature of GA1 in children.

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References

1 Hartley L, Khwaja O, Verity C. Glutaric aciduria type 1 and nonaccidental head injury. *Pediatrics* 2001; **107**: 174.
2 Bishop FS, Liu JK, McCall TD, Brockmeyer DL. Glutaric aciduria type 1 presenting as bilateral subdural hematomas mimicking nonaccidental trauma. *J. Neurosurg.* 2007; **106**: 222–6.
3 Hou LC, Veeravagu A, Hsu AR, Enns GM, Huhn SL. Glutaric acidemia type I: a neurosurgical perspective. *J. Neurosurg.* 2007; **107**: 167–72.
4 Piatt JH Jr, Frim D. Glutaric aciduria type 1 and nonaccidental head injury. *Pediatrics* 2002; **109**: 554.

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