Case Report

Glutaric Aciduria Type I: A Rare Metabolic Disorder Mimicking as Choreoathetoid Cerebral Palsy

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INTRODUCTION

Glutaric aciduria type I (GA I) is an autosomal recessive disorder of the degradation of lysine, hydroxylysine, and tryptophan, caused by a defect of the enzyme glutaryl-CoA dehydrogenase (GCDH).[1] The prevalence of GA I is 1 in 30,000. The number of diagnosed cases is, however, still low.[2] Enzyme deficiency results in an accumulation of glutaric and glutaconic acid, which can be measured by urine testing for organic acids. Glutaric acid has a cytotoxic effect and causes cerebral atrophy and brain damage.[3] This disorder is characterized by progressive extrapyramidal symptoms such as dystonia, dyskinesia, and choreoathetosis,[4] often misdiagnosed as athetoid cerebral palsy.[5]

CASE REPORT

A 7-year-old boy presented with choreoathetoid movements of both upper and lower limbs with dystonia and slurring of speech since 1½ years of age. He was born following uncomplicated pregnancy. There was no history of consanguinity among parents. There was no history of birth asphyxia or jaundice during infancy. He had a history of generalized tonic–clonic convulsion following a febrile illness at the age of 1½ year. The involuntary movements started following the episode of seizure. On examination, his head circumference was 58 cm (above the 97th centile), suggesting macrocephaly. He was conscious, well oriented and was having dysarthria. There was hypotonia in both upper and lower limbs with diminished reflexes. There was the presence of generalized choreoathetosis with dystonia of both upper and lower limbs.

Axial T2-weighted magnetic resonance imaging of the brain revealed bilateral frontotemporal atrophy with wide Sylvian fissure giving the “bat-wings” appearance [Figure 1]. Tandem mass spectrometry (MS) revealed increased levels of glutaryl carnitine (1.06 µmol/L; reference range – 0–0.41 µmol/L) and reduced levels of free carnitine (3.99 µmol/L; reference range – 9–65 µmol/L). Urine gas chromatography/MS showed increased excretion of glutaric acid, glutaconic acid, and 3-hydroxyglutaric acid. The diagnosis of GA I was confirmed on the basis of neuroimaging and biochemical studies. The patient was treated with riboflavin and carnitine along with protein-restricted diet especially diets lacking lysine and tryptophan. Follow-up after 3 months revealed some reduction in the involuntary movements.

ABSTRACT

Glutaric aciduria type I (GA I) is an autosomal recessive inborn error of metabolism caused by a deficiency of the enzyme glutaryl-CoA dehydrogenase. This disorder is characterized by progressive dystonia, choreoathetosis, and dyskinesia. It is often misdiagnosed as athetoid cerebral palsy. Laboratory evaluation usually demonstrates increased urinary excretion of glutaric acid and 3-hydroxyglutaric acid. We report a case of a 7-year-old boy presenting with choreoathetosis and dystonia, mimicking as choreoathetoid cerebral palsy. The presence of characteristic neuroimaging and biochemical studies led to the diagnosis of GA I.

KEYWORDS: Choreoathetosis, glutaric aciduria type I, macrocephaly

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The typical clinical feature and characteristic neuroimaging finding can give clue to this rare entity GA I, as seen in our patient. The delay in the diagnosis in our patient may be due to earlier misdiagnosis as athetoid cerebral palsy, which is very frequent in this disease. Early detection of this disease is very important, as diet restriction and riboflavin and carnitine therapy can limit and to some extent even reverse the neurological deficit. Features such as precipitation of a significant neurological disease by febrile illnesses, predominantly extrapyramidal manifestations, macrocephaly, and characteristic neuroimaging should arouse a suspicion of GA I.

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Conflicts of interest
There are no conflicts of interest.

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