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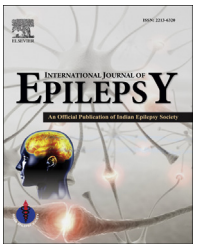

Epilepsy in inborn errors of metabolism: two cases with unusual presentation

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ABSTRACT

Inherited metabolic disorders are a rare cause of epilepsy in children. We describe a case of Glutaric aciduria type 1 presenting with West syndrome and a case of intermittent Maple syrup urine disease presenting with epileptic encephalopathy. Early diagnosis and institution of appropriate therapy may be life saving and may improve the long term neurodevelopmental outcome in children with inherited metabolic disorders.

1. Introduction

Inherited metabolic disorders are a rare cause of epilepsy.[1] However, in many metabolic disorders, seizures are the predominant symptom especially in newborns and infants e.g. pyridoxine dependent seizures, biotinidase deficiency and glucose transporter defect. It is most important to look for those inborn errors of metabolism which are treatable with supplementation of vitamins and cofactors or special diets. We report two unusual cases of inherited metabolic disorders with associated epilepsy.

2. Case 1

A 10-month-old male infant presented with developmental delay and jerky movements of the head and limbs since 6 months of age. He was the second child of non-consanguineous parentage. The antenatal and perinatal periods were uneventful. He achieved social smile at 3 months and partial neck holding at 5 months. Since 6 months of age, parents noticed jerky movements with flexion of the head and upper limbs, suggestive of flexor spasms. These movements would occur in clusters whenever the child woke up from

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sleep. The parents did not seek any treatment for these movements. The parents also noticed progressive stiffness of both the limbs and intermitted twisting postures of the hands and feet since 8 months of age. His vision and hearing were normal. There were no other seizure types. At the time of presentation, he had partial neck holding, was recognizing his mother and cooing. He had not started holding objects yet. His elder brother had died at 4 years of age due to pneumonia. He had been developmentally normal.

Examination revealed an alert infant with no facial dysmorphism or neurocutaneous features. His head circumference was 48 cm (>2 SD). His weight and length were age appropriate. The general physical and systemic examination revealed no abnormalities. Neurological examination revealed increased tone, brisk muscle stretch reflexes and bilateral extensor plantar responses. He also had hands and feet dyskinesias. The fundus examination was normal.

Investigations revealed normal hemogram, liver and kidney function tests. The EEG showed modified hypsarrhythmia (Fig. 1). The MRI of the brain showed hyperintense signal changes involving bilateral basal ganglia with fronto–temporal atrophy (Fig. 2). The tandem mass spectrometry of blood showed elevated glutarylcarnitine & low carnitine levels. His Urine gas chromatography and mass spectrometry showed highly elevated glutaric acid levels (>10,000 mmol/mol of creatinine).

A diagnosis of glutaric aciduria type 1 with West syndrome was made. The patient was started on riboflavin, carnitine and oral prednisolone (2 mg/kg/day). His spasms subsided within 5 days of starting steroids, which were tapered off after 2 weeks. The repeat EEG showed resolution of hypsarrhythmia. The patient was initiated with physical rehabilitation. Six months after the diagnosis, he has shown some improvement – he has achieved neck holding and has started reaching out for objects.

3. Case 2

An 18-month-old boy presented with history of jerky movements of the head along with regression of developmental milestones for the last 25 days. The patient was apparently well 25 days back when he developed fever and cough, which lasted for three days. On the third day of illness, the parents noticed jerky movements of the head with head flexion. These movements occurred multiple times in a day. There was no history of up-rolling of eyeballs or limb movements. The patient gradually lost the ability to walk, sit and talk over the next two weeks. He also stopped recognizing his parents. He also became drowsy and lethargic.

He had a similar episode at 13 months of age. During that episode, he had developed fever, cough and fast breathing. Then he developed jerky head movements and regression of milestones. He had been admitted in a private nursing home, where he was diagnosed to have pneumonia. He recovered completely and had regained his milestones within 7 days. He was the first child of non-consanguineous parentage. The antenatal and perinatal periods were uneventful. His developmental milestones were age appropriate prior to this illness. The family history was not significant.

On examination the child was lethargic but responsive. The anthropometry and vital parameters were normal. There was no facial dysmorphism. The general physical and systemic examination was normal. Neurological examination showed infrequent head myoclonic jerks, mild central hypotonia, normal muscle stretch reflexes and bilateral extensor plantar responses. There were no meningeal signs.

The MRI of the brain was normal. The EEG showed chaotic background with high voltage activity with intermixed spikes arising from bilateral leads (Fig. 3). There were also

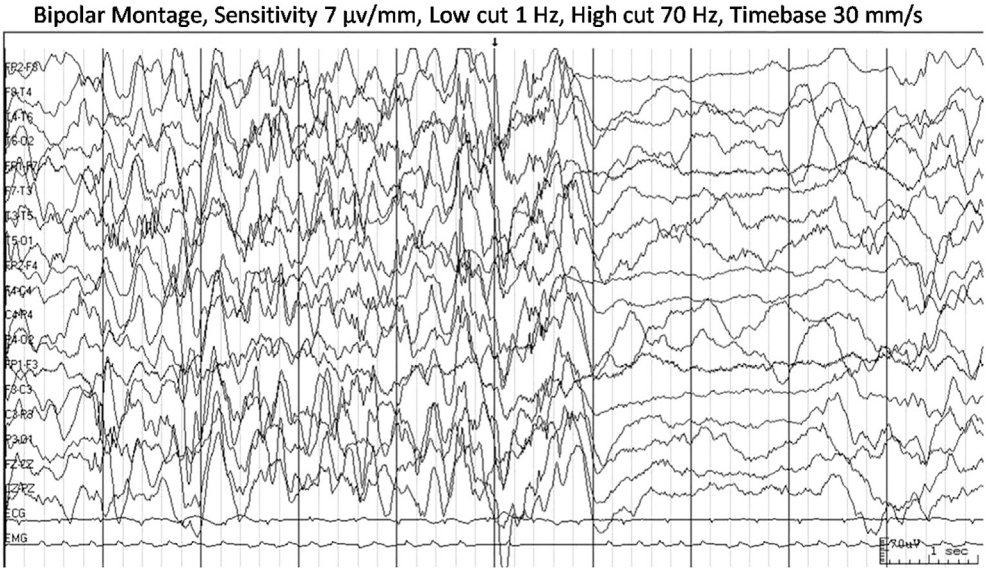

Fig. 1 – EEG of case 1: The sleep EEG shows a chaotic background with bilateral asynchronous high voltage delta activity with intermixed spikes and sharp waves followed by a period of electrodecrement. The pattern was suggestive of modified hypsarrhythmia.