

## Intermittent Ataxia with Early Onset Absence Epilepsy in Glucose Transporter Type 1 Deficiency Syndrome

A 4-year-old girl, born to healthy non consanguineous parents after an uneventful pregnancy and delivery with normal birth weight, presented with history of absence seizures and ataxia since 7 months of age. Absence seizures and ataxia were worse in the fasting state. She was otherwise able to walk and run, and had a normal speech. On examination, she had microcephaly (head circumference 46 cm); the tone was normal and deep tendon reflexes were brisk. She had mild ataxia.

A combination of early onset absence seizures with ataxia which were more prominent in fasting state led us to a suspicion of Glucose Transporter Type I (GLUT 1) Deficiency Syndrome. We considered other differential diagnoses like idiopathic early onset absence epilepsy, inborn errors of metabolism, episodic ataxia and cortical malformations. Cerebrospinal fluid (CSF) examination showed low glucose (30 mg/dL) compared to blood glucose (87mg/dL). Electroencephalography (EEG) showed generalized 2-3 Hz, 100-300 micro volts spikes, sharp waves and poly spike discharges. Arterial blood gas analysis, ammonia and tandem mass spectrometry were normal. Magnetic resonance imaging (MRI) of the brain was also normal. GLUT1 deficiency was confirmed with a missense mutation p.Thr295Met in exon 7 *SCL2A1* gene. The child was started on ketogenic diet following which the child became seizure free. The ataxia improved over a period of 4-5 weeks.

The classic phenotype in GLUT1 deficiency is infantile onset seizures, delayed neurological development and acquired microcephaly [1]. Cases with early onset absence epilepsy, intermittent ataxia, choreoathetosis, and dystonia and West syndrome have also been described [1,2]. The missense mutations are associated with mild to moderate clinical phenotypes [3]. There are few cases described in literature that share the same mutation as described in this child [4].

In summary, GLUT1 deficiency syndrome should be suspected in any child presenting with intermittent ataxia and early onset absence seizures which are more prominent in the fasting state. Ketogenic diet in these patients can prevent long-term morbidity.

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### REFERENCES

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