Positive impact of a modified Atkins diet on cognition, seizures control and abnormal movements in an adult with Glucose Transporter Type 1 Deficiency Syndrome

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Introduction

Glucose Transporter Type 1 Deficiency Syndrome (GLUT1DS) is a genetic disorder that impairs glucose transport across the blood-brain barrier. Patients may present with mild symptoms and remain undiagnosed or misdiagnosed with isolated seizure disorders, movement disorders or cognitive impairment while others could develop classic symptoms that lead to a rapid diagnosis. Seizures are the primary presenting symptom followed by abnormal movements and cognitive impairment.

GLUT1DS is most often an autosomal dominant disorder caused by a mutation in the SLC2A1 gene. Most cases are de novo rather than inherited mutations. Diagnosis is established by the presence of characteristic phenotypes, hypoglycemic crisis and a mutation in the SLC2A1 gene. There is no cure and therefore, strategies for providing an alternative energy source to the brain are essential. Ketogenic diet therapies (KDT) are high fat, low carbohydrate diets that induce fat metabolism and ketone body production. KDT reduces seizures and abnormal movements but its impact on cognition remains unclear. The classic KD is strict with regard to precisely weighing proportions of foods using a gram scale, and it is therefore difficult to follow for adolescents and adults. The modified Atkins diet (MAD), a less strict diet, can be an alternative in this population.

Objective

We present a case of a woman diagnosed with GLUT1DS as an adult who, following the initiation of MAD, not only demonstrated improvement in the frequency and severity of abnormal movements and seizures but also displayed significant improvement in cognitive function resulting in independence in performing activities of daily living (ADLs).

Case Presentation

The patient was born full-term by vaginal delivery following an uncomplicated pregnancy. Family history was negative for genetic disorders and consanguinity.

Her early development was normal until age ten months, when she developed mild language regression and afebrile generalized tonic-clonic seizures.

She demonstrated slowed progression in cognitive, language, and motor skills.

At 19 months old, she experienced episodes with sweaty palms, difficulty walking, and dystonic movements with right arm and leg inversion or extension, neck contraction, and head rotation followed by rhythmic, circular oscillating movements of the head. She also developed absence seizures.

Her tonic-clonic seizures were controlled in infancy, but absence seizures, dystonic episodes, language impairment, and severe intellectual disability persisted into adulthood.

At 22 years old, her genetic evaluation showed a missense mutation in the SER131TYR in exon 7 of the GLUT1 gene.

At 26 years old, she started restricting carbohydrates to 30 grams per day, but she initially did not adhere strictly to the diet.

At 27 years old, she was counseled at the Johns Hopkins Adult Epilepsy Diet Center and she adhered strictly to MAD. Her dystonic episodes and seizures improved significantly.

Discussion

This case report highlights the favorable impact of KDT on cognitive function, seizure control, and reduction in abnormal movements in an adult newly diagnosed with GLUT1DS, the effectiveness of MAD as an alternative to the classic KD, and its favorable safety profile.

MAD may be preferred in adults and adolescents as a more liberal option than the classic KD which may result in higher adherence rates.

The patient had seizure resolution in early adulthood without recurrence after tapering antiseizure drugs, and she has remained on diet monotherapy since then.

Our patient had daily dystonic episodes since childhood and, even despite her late diagnosis and treatment, the frequency of her spells significantly decreased from daily to every one or two months when she started MAD.

The efficacy of KDT on improving cognitive function has been poorly studied and data in adults is almost nonexistent.

She can now perform ADLs independently with verbal cues including showering, speaking in complete sentences, understand instructions from her parents and caregivers, and is engaged in reading lessons.

The adverse effects of KDT are common but easy to manage. Nephrolithiasis, dyslipidemia, gastrointestinal side effects, weight loss, and amenorrhea are among the most frequent side effects of the diet.

Conclusion

This case report demonstrates the efficacy of MAD in the treatment of an adult newly diagnosed with GLUT1DS and its significant effect on cognitive function, maintained seizure control, reduction in movement disorder symptoms and ultimately, improvement in quality of life.

References