Glucose transporter type 1 (GLUT-1) Deficiency Syndrome – delayed diagnosis and treatment: case report

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Glucose transporter type 1 deficiency syndrome (GLUT1-DS) is a treatable metabolic disorder caused by a mutation in SLC2A1 gene. The most severe classic phenotype comprises infantile-onset epileptic encephalopathy associated with delayed development, acquired microcephaly, motor coordination disturbances, and spasticity. There are also observed the less severe clinical features, associated with paroxysmal exercise-induced dystonia with or without epileptic seizures. We describe the woman, who was diagnosed as epileptic patient and treated for years with different antiepileptic drugs with no clinical effect. She had the only two generalized tonic clonic seizures in her life. Instead of them she suffered from increasing frequency of the paroxysmal involuntary movements of lower limbs, leading to gait disturbances and falls, which were wrongly diagnosed as epileptic seizures, too. The jerks of the head and limbs were observed for the first months of her life. The symptoms were provoked by stress and exertion. Additionally, slight mental retardation was observed during her growth. Due to similarity of seizures symptomatology as well as paroxysmal dystonic movements, related to them status epilepticus and dystonic status, clinical differentiation may be difficult. The lumbar puncture was performed, then low glucose concentrations in cerebrospinal fluid was set. The results of genetic tests revealed the missense mutation of protein p.Arg333Trp, and confirmed the diagnosis of GLUT-1-DS1 syndrome. Delayed diagnosis caused the great problems with acceptance of the ketogenic diet, which is suggested as the treatment of choice in GLUT-1 deficiency syndrome.