MILD ADOLESCENT/ADULT ONSET EPILEPSY AND PAROXYSMAL EXERCISE-INDUCED DYSKINESIA DUE TO GLUT1 DEFICIENCY

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Purpose: Paroxysmal exercise induced dyskinesia (PED) and epilepsy without intellectual disability have recently been recognized as manifestations of deficiency of the glucose transporter GLUT1, due to mutations in the gene SLC2A1. We describe clinical features of an Israeli family with this disorder.
Method: The family was ascertained and clinically assessed as part of a systematic study of the genetics of epilepsy in Israel. SLC2A1 was examined by direct sequencing.
Results: This Ashkenazi Jewish family had 6 definitely affected members in 2 generations with an autosomal dominant mode of inheritance. Two had PED, three had epilepsy and one had both. A seventh member appeared to be a phenocopy with probable benign partial epilepsy of childhood. Seizures began at a mean age of 19 years (range 13-30 years), they were tonic-clonic in 3 and myoclonic in 1 in the context of a syndromic diagnosis of probable idiopathic generalized epilepsy. The mean age of dyskinesia onset was 13 years (range 10-15 years), with symptoms predominantly experienced in the legs after walking or exercises, with a cramping sensation and dystonic movements of toes and feet. A missense mutation in SLC2A1 (c.950A>C; p.N317T) was detected in five living affected members, but absent in 2 non-affected first degree members and in the subject believed to be a phenocopy.
Conclusion: The clinical picture of mild epilepsy with onset in adolescence or early adulthood plus PED, which is easily overlooked, should raise suspicion of GLUT1 deficiency.