Abstract

Neuroacanthocytosis syndromes are a group of heterogeneous neurodegenerative disorders characterized by neurological abnormalities associated with acanthocytosis. Chorea-acanthocytosis is one of entity of neuroacanthocytosis syndrome caused by the mutations of the VPS 13A gene which encoding for chorein protein. Here we report a case of 37 year old male presented with increasing involuntarily movements following seizures with gait disturbance and dysarthria. Neurological examination revealed he had choreiform movements, orofacial dyskinesias, feeding distonia. His peripheral blood smears revealed acanthocytosis of 70% and elevated creatinine kinase of 817 U/L. Peculiar features of orofacial dyskinesias, feeding dystonia and self lip mutilation favored the diagnosis of chorea-acanthocytosis. VPS13A gene test or western blot for chorein can be used to confirm the diagnosis.