Consciousness disturbances in megalencephalic leukoencephalopathy with subcortical cysts.

Megalencephalic leukoencephalopathy with subcortical cysts (MLC) is a genetic disorder featuring diffuse MRI white matter abnormalities and a discrepantly mild clinical picture. It is related to different mutations in MLC1 gene encoding a putative membrane protein of still unknown function. We report on a genetically proven MLC patient who presented with a peculiar clinical course characterized by a prolonged comatose state following a minor head trauma at 12 years of age. The disturbance of consciousness lasted for over four months and then gradually improved. Proton MR spectroscopic imaging studies showed a moderately severe depletion of N-acetylaspartate restricted to the white matter with sparing of the cortical grey matter. The full recovery from coma suggests a transitory functional impairment of the structures implicated in the maintenance of consciousness.