Hereditary diffuse leukoencephalopathy with spheroids (HDLS) is a rare progressive white matter disease with a wide range of clinical symptoms, including dementia, behavioral changes, seizures, pyramidal signs, ataxia, and parkinsonism.

Recently, mutations in the colony-stimulating factor 1 receptor gene (CSF-1R) were identified as the genetic cause of HDLS. We report the case of a 38 years old man, presenting with cognitive and behavioral symptoms, that developed rapidly progressive generalized dystonia and parkinsonism. A frameshift deletion in CSF-1R has been identified in this patient and his 80-years mother featuring only bipolar disorder.