

Can miglustat halt the progression of Niemann-Pick disease type C

J. Kim

Neurology, Dong-A University Hospital, South Korea

Miglustat has been known the only medical treatment to improve or halt the neurological symptoms in Niemann-Pick type C (NP-C). The efficacy of miglustat was evaluated for the first two Korean patients. Case 1 developed delusion and abnormal posturing on both hands at the age of 18. All symptoms were gradually aggravated. At 24, he became completely dependent on caregivers and wheelchair bound. Case 2 was the younger sister of case 1. Her symptoms began with dystonia in the right hand at 19 years old and were milder than her brother. Both patients were evaluated for dystonia and ataxia using the Dystonia Movement Scale (DMS, maximum score 120) and Scale for the assessment and rating of ataxia (SARA, maximum score 37) before and 18 months after the administration of miglustat. 600mg of miglustat a day was given to both patients. The baseline scores of DMS were 74 and 29 for case 1 and 2, respectively. They changed to 96 and 27 after the administration of miglustat for 18 months. The baseline scores of SARA were 26.5 and 13. They changed to 39 and 14. After miglustat therapy, the case 1 worsened for both dystonia and ataxia. However, the case 2 demonstrated a stabilization of her symptoms. Our results show that miglustat treatment can halt the disease progression at least for a period of time in the early stage of the illness. The earlier detection of the condition seemed to be important for the better efficacy of miglustat in NP-C.