Dopamine transporter image in Niemen-pick disease type c

J. Kim[1], S. Y. Lee
[1]Neurology, Dong-A University Hospital, South Korea

Niemann-Pick disease type C (NP-C) is a rare autosomal recessively inherited lysosomal storage disorder characterized by progressive neurological symptoms and various degrees of visceral involvement.The patient was a 24-yr-old male presented with psychosis, abnormal posturing, and gait disturbance. His birth and development had been unremarkable. At the age of 18, He started to develop delusion and abnormal posturing on both hands. At 21, He had to be hospitalized in department of psychiatry because of troublesome psychosis such as visual and auditory hallucinations, aggressive and impulsive behavior. He also developed gait disturbance and cognitive impairment around that time. Despite of symptomatic treatment, all symptoms were gradually aggravated, so at the age of 24, he became completely dependent on caregivers and wheelchair bound. Family history revealed that his younger sister had similar symptoms. On neurologic examination, He showed generalized dystonia that is prominent in both upper limbs, severe ataxic gait, VSGP and severe dysarthria. Brain Magnetic resonance image (MRI) showed no definite signal change, but mild atrophy of posterior part of brain was seen and ^{18}F-FP-CIT positron emission tomography (PET) scan showed mildly decreased uptake in right caudate and anterior putamen. NPC1 gene sequencing revealed a compound heterozygote mutation which was already known as a genetic cause of NP-C, one in exon 9 (c.1552CT [R518W]) and one in exon 18 (c.2780CT [A927V]). Filipin staining test of cultured fibroblasts from skin, which is another key diagnostic test for NP-C, was done and the result was positive. There has been no systematic study in NP-C using ^{18}F-FP-CIT scan that demonstrate presynaptic dopaminergic neuronal loss. Our case had decreased dopaminergic uptake in ^{18}F-FP-CIT scan, but the pattern was much differ from parkinson's disease, in which deficit is predominantly in dorsal, posterior putamen.