Annual Congress on

Advancements in Neurology, Neuroscience and Pediatric Neurology

June 18-19, 2018 Rome, Italy

A novel gene mutation in PANK2 in a patient with severe jaw-opening dystonia

Zuhal Yapici
Istanbul University, Turkey

Pantothenate Kinase-Associated Neurodegeneration (PKAN) is a rare neurodegenerative condition. Major clinical features include progressive dystonia, pigmentary retinopathy, spasticity and cognitive decline. The typical MRI sign of the disease, known as "eye-of-the-tiger", is what makes differential diagnosis possible. We here describe a 16-year-old male patient with PKAN presenting with severe and sustained jaw-opening dystonia which may be due to heterogeneous etiologies showing poor response to treatment. Herein, long-term follow-up and genetic results of a PKAN case who experienced severe jaw-opening dystonia are presented and discussed.

yapicizuhal@gmail.com