Original article

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Long-term follow-up of two siblings with adult-onset neuronal ceroid lipofuscinosis, Kufs type A

Çiğdem Özkara [1], Ayşegül Gündüz [1], Tülin Coşkun [1], Bengi Gül Alpaslan [1], Burcu Zeydan [1], Şakir Delil [2], Mikko Muona [3], Anna-Elina Lehesjoki [4], Meral E. Kızıltan [1]

[1] Istanbul University, Cerrahpasa School of Medicine, Department of Neurology, Istanbul
[2] Batı Bahat Hospital, Istanbul, Turkey
[3] Institute for Molecular Medicine Finland, University of Helsinki, Helsinki ; Folkhälsan Institute of Genetics, Helsinki ; Research Programs Unit, Molecular Neurology and Neuroscience Center, University of Helsinki, Helsinki
[4] Folkhälsan Institute of Genetics, Helsinki Finland; Research Programs Unit, Molecular Neurology and Neuroscience Center, University of Helsinki, Helsinki, Finland

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- Arsov et al. (2011) identified patients with Kufs A disease with *CLN6* mutations, and most patients exhibited myoclonus, seizures, and dementia, followed by ataxia at around 30 years of age, without any loss of vision.

- However, neither our patients nor their relatives had dementia or extrapyramidal findings.

- Electrophysiological recordings of myoclonus or indirect electrophysiological indicators of increased cortical activation, such as the presence of the C reflex, did not show changes despite adequate control of seizures.

- However, our patients provide evidence for the inhibition of various brainstem reflexes in NCL and establish that these abnormal findings deteriorate with time.

- Persistent photosensitivity is an important clinical feature in NCL associated with *CLN6* mutation.

- Piracetam provided dramatic relief of myoclonic seizures and ataxia in the presented cases. Piracetam may be considered in the palliative treatment of these patients.