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Correspondence

Reply to: Double trouble progressive external ophthalmoplegia and Huntington's disease

Keywords:
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We read the letter by Finsterer and Zarrouk-Mahjoubb [1] about our paper "A novel mitochondrial tRNA(Ala) gene variant causes chronic progressive external ophthalmoplegia in a patient with Huntington disease" [2].

We thank the colleagues for manifesting interest in our paper and expressing their opinion.

However, we do not agree on the considerations in respect to the clinical condition affecting our patient.

From a clinical point of view, the patient presents a myopathic picture characterized by bilateral ophthalmoparesis, dysphagia, dysphonia and mild proximal limb weakness. It then associates numbness and bilateral deafness. About 10 years later, extrapyramidal signs such as abnormal involuntary movements involving the head and limbs, imbalance and cognitive decline appeared.

It is well known that late-onset Huntington disease usually present with a 60–79 year onset range and a CAG expansion size ranging from 38 to 44 repeats, or even less. These patients develop chorea and motor, cognitive and psychiatric symptoms [3,4,5].

Given the occurrence of a 38 CAG pathological expansion in the *HTT* gene in our 70-year-old patient, all clinical symptoms cannot be attributed with certainty only to mitochondrial mutation and a "double trouble", a concomitant action between the two clinical conditions, which is what we have described in the paper, remains the more likely explanation.

Yours sincerely,

References

[1] J. Finsterer, S. Zarrouk-Mahjoubb, Double trouble progressive external ophthalmoplegia and Huntington's disease, Mol. Genet. Metab. Rep. 7 (2016) 77.
[2] M. Filosto, G. Lanzi, C. Nesti, V. Vielmi, E. Marchina, A. Galvagni, S. Giliani, F.M. Santorelli, A. Padovani, A novel mitochondrial tRNA(Ala) gene variant causes chronic progressive external ophthalmoplegia in a patient with Huntington disease, Mol. Genet. Metab. Rep. 6 (2016) 70–73.
[3] P.J. Garcia-Ruiz, J. Garcia-Caldentey, C. Feliz, J. Del Val, A. Herranz, J.C. Martínez-Castrillo, Late onset Huntington's disease with 29 CAG repeat expansion, J. Neurol. Sci. 363 (2016) 114–115.
[4] M.R. Cornejo-Olivas, M.A. Inca-Martinez, K. Espinoza-Huertas, D. Veliz-Otani, M.R. Velit-Salazar, V. Marca, O. Ortega, I.F. Cornejo-Herrera, S. Lindo-Samanamud, P. Mora-Alferez, P. Mazzetti, Clinical and molecular features of late onset Huntington disease in a Peruvian cohort, J. Huntingtons Dis. 4 (2015) 99–105.
[5] H. Lipe, T. Bird, Late Onset Huntington Disease: clinical and genetic characteristics of 34 cases, J Neurol Sci 276 (2009) 159–162.

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