Open Science Journal of Psychology
2016; 3(2): 9-11
http://www.openscienceonline.com/journal/osjp

Atypical Psychotic Symptoms in Lafora´s Disease: A Case Report

Núñez Domínguez L. A.[1, *], Reclusa Martín P.[2], Martínez Olea A.[2], Burgos Martínez N.[2], Uriz Sanjosé G.[2], Mayoral González M.[2], Olorón Tolosa A.[2]

[1]General Practiconer, Centro Infanta Elena, Cordovilla, Navarra

[2]Centro Infanta Elena, Cordovilla, Navarra

Email address

medico.infantaelena@sarquavitae.es (Núñez D. L. A.)

[*]Corresponding author

To cite this article

Núñez Domínguez L. A., Reclusa Martín P., Martínez Olea A., Burgos Martínez N., Uriz Sanjosé G., Mayoral González M., Olorón Tolosa A. Atypical Psychotic Symptoms in Lafora´s Disease: A Case Report. *Open Science Journal of Psychology.* Vol. 3, No. 2, 2016, pp. 9-11.

Received: August 8, 2016; **Accepted**: August 19, 2016; **Published**: September 2, 2016

Abstract

Lafora´s disease is a strange type of myoclonic disorder (there have been described no more than 200 cases in the literature), with genetic basis. Some mutations provoke a beginning in the adolescence and a quickly development. At present it doesn't exist a treatment, none to the disease, but also to the different neurologic associated disorders (myoclonus, seizures, dementia) and fatal outcome. Along the evolution, it´s usual to find psychotic symptoms, especially auditory hallucinations. We present a case with atypical evolution: The subject presents auditory hallucinations, with a good response to psychopharmacological treatment. The presence of these symptoms could be observed in the first years of the diseases. We describe the status of the patient at the admission, the evolution during the stay, with the increase of symptoms in several areas (cognitive, motor, neurologic), and the current status, after hospitalization, where the patient begun receiving enteral nutrition by gastrostomie, due to the limitations to receive the treatment and /or nutrition by oral way. This case is also interesting due to the use of Metformine, an antidiabetic agent, whose use has been suggested as therapeutic alternative for this syndrome.

Keywords

Lafora Disease, Myoclonus, Treatment, Dementia, Psychosis, Visual Hallucinations

1. Introduction

Lafora disease is a very strange kind of progressive mioclonic epilepsy, with recessive autosomic transmission, with higher incidence at the countries round the Mediterranean Sea. The disease typically debuts at adolescence, with quick progression until global deterioration with generalized epilepsy until death, which commonly occurs about 10 years after the appearance of first symptoms [1, 2].

The diagnoses are made through the analysis of clinical data, including epileptic seizures, and myoclonias. Genetic studies typically show several mutations in EPM2A o EPM2B genes, two of the more usual genes associated with the disease [3]. Skin biopsy shows different anatompathological changes compatible with the diagnoses of Lafora disease [4].

The presence of psychotic symptoms, basically visual hallucinations, It´s very common [5, 6].

We describe a case of Lafora disease that presents visual and auditory hallucinations, and delusions of reference.

2. Case Report

Male, 24 years old, joined the Centro de Atención Integral a la Discapacidad (CAIDIS) Infanta Elena de Cordovilla (Pamplona, Navarra) in 2010, due to the impossibility to be cared after at home. He was diagnosed of Lafora Disease in 2009 (17 years old). He presented medical problems at the age of 13. He was born in Moldavia, Romania, without familiar antecedents of epilepsy nor neurologic disease.

A skin biopsy was performed to confirm the diagnosis: the results were some anatomopathological changes in the skin

(Lafora bodies). It also was done a genetic study; it was found a heterozigotic mutation in EPM2A gene.

IQ was normal until the patient was 13 years old. The first manifestations of the disease occur at that age. He had just finished college with severe difficulties at the age of 16. At that moment, the progression of the disease limited him very seriously and he stopped his studies.

In addition, the medical record shows a large history of admittances at Emergency Department, due to epileptic seizures and behavioral disorders (heteroagresivity at home), related to the patient refuse to follow the treatment. It is recorded that once he even slept with a knife under the pillow.

According to the medical records of that time, he presented a moderate cognitive deficit, myoclonia, more important in the arms, and epileptic seizures, with bad response to the treatment (up to 7 antiepileptic drugs at the same time). Additionally, he presented visual hallucinations, with good response to antipsychotic treatment.

2.1. Admission in CAIDIS Infanta Elena

In the moment of the admittance, the patient showed a moderate functional dependence for the basic daily activities (Barthel´s Index 45), though he maintained the capacity to eat by himself. He could walk and perform the transferences manually. He was also continent.

The main problem for personal care was the variability in this behavior: he presented several episodes of aggressiveness against the staff. Moreover, although he was able in some moments to collaborate with dressing and cleaning, in other moments he was absolutely dependent. It was also necessary to adapt the ambiance due to the presence of hallucinations and behaviors associated with them.

2.2. Evolution During the Admission

During the stay in the CAIDIS, we observe an increase of blocks into the process of food consumption (he doesn't open the mouth, nor manages to cud,…), with the result of a progressive decrease of the quantity of food consumed. Besides, the speech therapist detects the beginning of archaic reflex, with great difficulty for food administration. The use of technical helps for the administration of food and drinks are needed.

The physiotherapists find a very important handicap for the application of their treatment in the sense of the great variability in the physical and/or psychological status of the patient. They observe important loss of coordination and balance, very usual in the evolution in this disease, together with the maintenance of physical strength

The neuropsychological evaluation was conditioned by the important cognitive decline: it was not possible to run the standard tests, being necessary to evaluate the patient through observation of his behavior: we detect reduced language and a decrease in the speed of process, together with a increase in the latency of answers up to 15-30 seconds. He maintains the capacity to express feelings and demands, with partial conscience of his handicap. On the other side, he has important limitation in the level of comprehension, getting to respond to simple orders and clear verbal language. Finally, a decline in the maintenance of written language is observed. The use of several group session based techniques of cognitive stimulation and behavior therapy twice a week, as well as the use of Neuron Up program, with analogous frequency has generated minimal and ineffective results, due to disease evolution.

In December 2015, the staff referred the presence of visual (sudden stare to a fixed point and screams such as:" Get out, Get out of the room") and auditory hallucinations ("Shut up"), associated with behavioral disorders (Knocking on the wall, the doors and the electrical sockets, where he thought the voices came from), with the result of some wounds in his feet. Oral risperidone (up to 2 mg/day) was associated, with a progressive remission of the psychotic symptoms and the behavioral disorders. However, several delusions of reference appeared (the patient thought that the staff was trying to poison him), leading to a refuse of both treatment and food. For this reason treatment is revised, beginning with the injection of Paliperidone Palmitate (up to 100 mg per month) as only way to assure compliance. We observe a good tolerability and symptomatic response, but together with an increase in the number of epileptic seizures and important loss of weight. Due to the last, the patient was derived to general Hospital for gastroenteric feeding.

2.3. Current Symptomatology

The patient remains bed ridden, with little activity and few reactions to stimulus, probably due to physical weakness. Over time he is increasing both body mass and physical tone, with a low level of epileptic seizures. Psychotic symptoms have re-appeared, which has led to restarting the treatment with Paliperidone Palmitate, up to 75 mg per month.

Disease evolution can be seen in speech and language of the patient. At present, we observe that the patient shows faltering oral emission, gestual apraxia, altered coordination and non fluent language, though maintaining structure and comprehension of both oral and written language.

On the other side, patient presents a severe cognitive deterioration, yet he is occasionally able to maintain the contact with environment and makes attempts to communicate verbally. We observe some behaviors associated to psychotic symptoms, such as refuse to treatment, physical aggressiveness against the staff or another residents and hallucinatory nature ideation, which lead to a severe irritability status.

He needs technical aids, such as a tilting wheel chair with floating head and pelvic and shoulder fastening. He obtained a score of zero in Barthel´s Index, presenting total dependence in basic daily living activities. In addition he presents daily epileptic seizures and short-term myoclonias.

Several adjustments in the antiepileptic pharmacological treatment have been carried out, with the result of a decrease in both frequency (to just weekly) and intensity of the seizures. We have added Metformine, at a dose of 850 mg twice a day.

3. Discussion

The characteristics shown by the patient in this case are the usual in this disease: beginning in adolescence, progressive physical and psychological deficits, bad response to psychopharmacological treatments to control epileptic seizures and presence of psychotic symptoms [7, 8].

What makes this case peculiar is the presence of auditory hallucinations and delirious ideation, both non habitual symptoms as we noted in a wide review of the literarure [9]. In one hand, auditory hallucinations are very strange in cognitive disorders; in the other hand, the presence of prejudice delirium is very common in the same disorders, but with no response to psychopharmacological treatment; in the present case, the response is very good.

We also consider very interesting this case because it has been possible the use of Metformine, antidiabetic agent, which has been proposed as an alternative therapy in this disease after showing efficacy in animal models [10]

We finally think that description and diffusion to scientific community of this singular case may help to a better understanding of this disease and to a more complete caring of patients with very rare disorders.

Acknowledgements

We would like to thanks Iñaki Asensio for the revision of the final version of the manuscript

References

[1] Delgado-Escueta AV. Advances in Lafora progressive myoclonus epilepsy. Curr Neurol Neurosci Rep2007; 7: 428–433.

[2] Monaghan TS, Delanty N. Lafora disease: epidemiology, pathophysiology and management. CNS Drugs 2010; 24: 549–561.

[3] Minassian BA. Lafora's disease: towards a clinical, pathologic, and molecular synthesis. Pediatr Neuro 2001; 25(1): 21-9.

[4] Serratosa JM, Gomez-Garre P, GallardoME, Anta B, de Bernabe DBet al. A novel protein tyrosine phosphatase gene is mutated in progressive myoclonus epilepsy of the Lafora type (EPM2). HumMol Genet 1999; 8: 345–352.

[5] El Tahry R, de Tourtchaninoff M, Vrielynck P, Van Rijckevorsel K. Lafora disease: psychiatric manifestations, cognitive decline, and visual hallucinations. Acta Neurol Belg 2015; 115(3): 471-4.

[6] Andrade DM, del Campo JM, Moro E, Minassian BA, Wennberg RA. Nonepileptic visual hallucinations in Lafora disease. Neurology 2005; 64(7): 1311-2.

[7] Jansen AC, Andermann E. Progressive Myoclonus Epilepsy, Lafora Type. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Fong CT, Mefford HC, Smith RJH, Stephens K, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2016.2007 Dec 28 [updated 2015 Jan 22].

[8] Striano P, Zara F, Turnbull J, Girard JM, Ackerley CA, Cervasio MR, et al. Typical progression of myoclonic epilepsy of the Lafora type: a case report. Nat Clin Pract Neurol. 2008; 4(2): 106-10.

[9] Turnbull J, Girard JM, Lohi H, Chan EM, Wang P, Tiberia E, et al. Early-onset Lafora body disease. Brain 2012; 135(Pt 9): 2684-98.

[10] Berthier A, Payá M, García-Cabrero AM, Ballester MI, Heredia M, Serratosa JM, et al. Pharmacological Interventions to Ameliorate Neuropathological Symptoms in a Mouse Model of Lafora Disease. Mol Neurobiol 2016; 53(2): 1296-309.