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LETTER TO THE EDITOR

Paroxysmal tonic upgaze with epileptiform discharges and calcarine sulcus lesion

Desviación paroxística de la mirada en supraversión con descargas epileptiformes y lesión en cisura calcarina

Dear Editor:

Benign paroxysmal tonic upgaze syndrome was first described in 1988.[1–3] It is a type of episodic dystonia characterised by an upgaze movement preserving the horizontal visual axis, with normal level of consciousness, normal findings in the neurological examination, and unknown aetiology. Familial cases have been reported, and mutations in the *CACNA1A*,[4] *GRID2*,[5] and *SEPSECS* genes, and chromosome 15 have been identified (both in cases of maternal deletion, such as in Angelman syndrome, or paternal deletion, for example in Prader-Willi syndrome).[6] Clinical onset occurs in the first months of life, and symptoms disappear during sleep and tend to recover spontaneously, although secondary forms must be ruled out.[1,3,7] This group of signs and symptoms represents a diagnostic challenge for clinicians due to the broad differential diagnosis, including a movement disorder manifesting as dystonia, seizures, or non-epileptic paroxysmal disorders, with video encephalography (EEG) being a support tool for diagnosis.[6] EEG findings are normal in the majority of cases, although some show epileptiform discharges in the occipital region.[1,7] Brain imaging studies yield normal results, with the exception of a few cases showing some lesions in the visual pathway.[3]

We report the case of a young woman with a history of rheumatoid arthritis, which was appropriately controlled with methotrexate and non-steroidal anti-inflammatory drugs, and Graves disease with no thyroid orbitopathy, under treatment with 5 mg thiamazole daily. She presented sudden episodes of involuntary, conjugate eye deviation in the upper vertical axis with preserved consciousness, which had not manifested during her early childhood. These episodes lasted seconds and had manifested with an inconstant frequency over the past 6 months. She reported no other symptoms. Neurological examination findings between episodes were normal, and no relatives were affected. Blood analysis revealed unaltered thyroid function and no exacerbation of connectivopathy or heavy metal accumulation.

The ophthalmic examination yielded normal results. No relevant results were obtained in the echocardiography or 24-h Holter monitoring study.

Suspecting an epileptic aetiology due to the stereotyped nature of the episodes, we requested an EEG study, which revealed focal interictal activity with spikes in the right posterior quadrant (P4-T8 electrodes) (Fig. 1). In the brain MRI scan, T2-weighted and FLAIR sequences showed a cortical hyperintensity in the primary visual cortex (Fig. 2), corresponding to the calcarine sulcus (Brodmann area 17). The calcarine sulcus divides the medial surface of the occipital lobe and delimits the location of the primary visual cortex.[8] No pathological enhancement was observed after contrast administration.

The patient started monotherapy with 500 mg levetiracetam every 12 h for 3 months, with no change in the frequency of episodes.

Occipital epilepsy represents 5%–10% of focal epilepsies. It is characterised by short seizures that propagate rapidly. The auras of these seizures include simple visual symptoms (which may be negative [eg, scotomas or hemianopias] or positive [eg, dysphotopsia]), complex visual hallucinations, autonomic symptoms (as in the case of benign occipital epilepsy or Panayiotopoulos syndrome), and eye movement sensation.[9,10] Discharges in the posterior cortex may propagate to the temporal lobe, causing the corresponding symptoms, or to the suprasylvian convexity or medial surface from the supracalcarine area, mimicking symptoms of frontal or parietal seizures.[9–12]

Our patient presented symptoms consistent with occipital lobe epilepsy manifesting as eye movement and a lesion at the occipital junction.

In our case, complementary test findings (EEG and brain MRI) supported the epileptic aetiology, although after 3 months, antiepileptic monotherapy did not modify the frequency or characteristics of episodes. No genetic study has been requested to date; however, in patients whose relatives present similar symptoms or such signs as paroxysmal torticollis of infancy, episodic ataxia, hemiplegic

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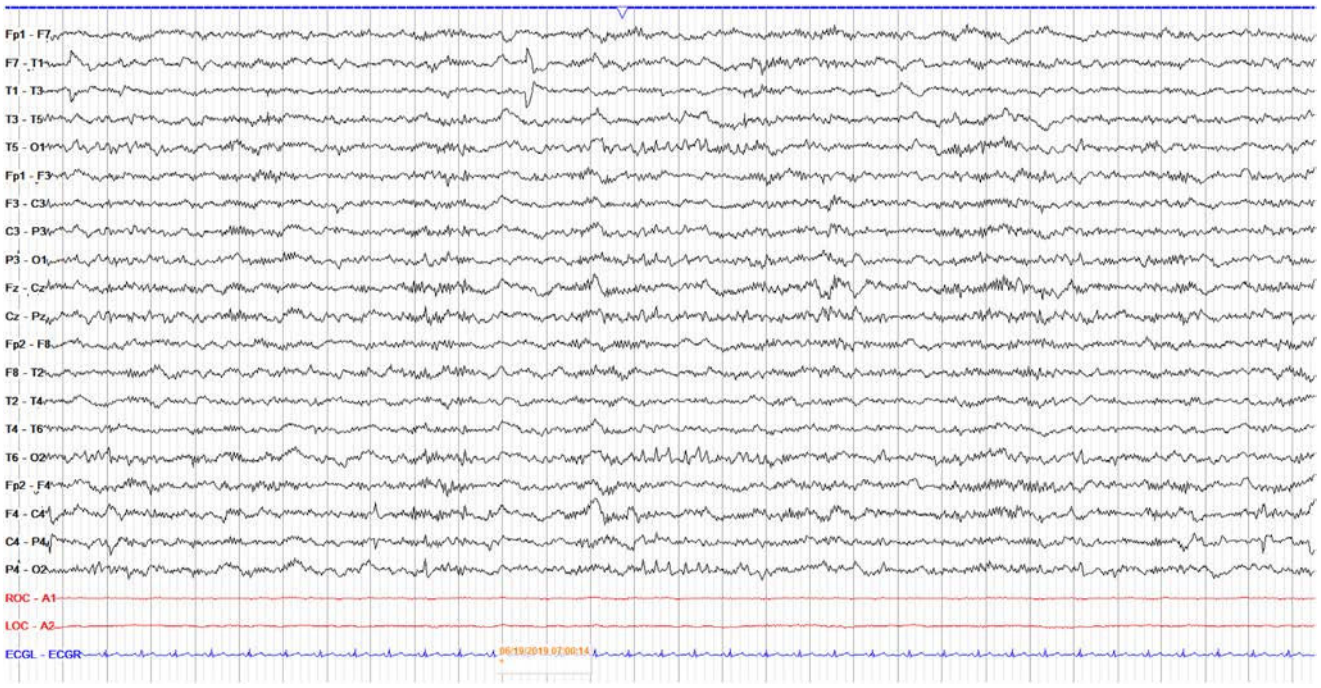

Figure 1 Encephalography showing paroxysmal activity in the form of spikes in the right posterior quadrant (P4 > O2 > T6).

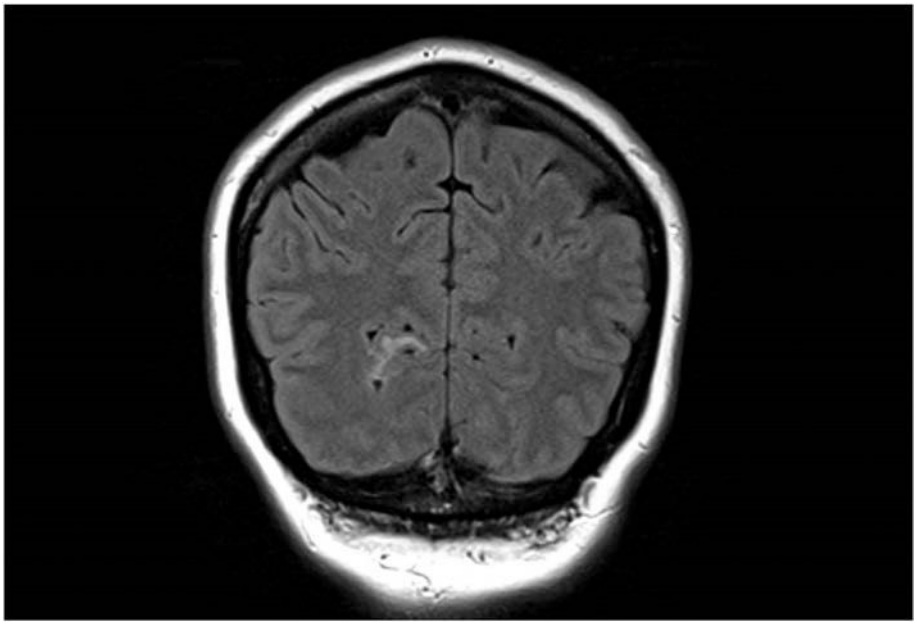

Figure 2 Coronal FLAIR MRI sequence showing a hyperintense lesion in the primary visual area.

migraine, or even arrhythmia suggestive of channelopathy, it is necessary to identify *CACNA1* mutations[4] and to consider the other genes involved.

To conclude, we underscore the interest in the electrical activity and brain imaging findings observed in this case. Their relevance resides in the importance of familiarising ourselves with such an infrequent syndrome as conjugate upward deviation of the eyes, as well as expanding the differential diagnosis by considering focal occipital epilepsy, in which video EEG may be used to monitor episodes. Our aim must be to avoid unnecessary treatments and to determine the genetic relationship between neurological and heart diseases.

Funding

None.

The patient has signed an informed consent form.

This study complies with the ethical considerations of our hospital's ethics committee.

Conflicts of interest

None.

Appendix A. Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.neurop.2025.100185.

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