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Cerebral visual impairment in a patient with Charcot-Marie-Tooth syndrome and Neurofibromatosis type 2: an uncommon case report

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BACKGROUND

Charcot-Marie-Tooth disease (CMT) is a hereditary neurological disorder primarily affecting the peripheral nerves, leading to progressive muscle weakness, atrophy, and sensory loss in the extremities. The CMT1B subtype is caused by mutations in the MPZ gene, whose expression is restricted to Schwann cells and is absent in the central nervous system (CNS). No reports have identified cognitive or cerebral visual impairment in CMT1B. Neurofibromatosis type 2 (NF2) is a rare genetic disorder characterized by the formation of multiple schwannomas. Visual impairment is not rare in NF2 and is most often due to optic nerve sheath meningiomas, compression of the optic pathways by CNS tumors, or even retinal abnormalities and cataracts.

CASE PRESENTATION

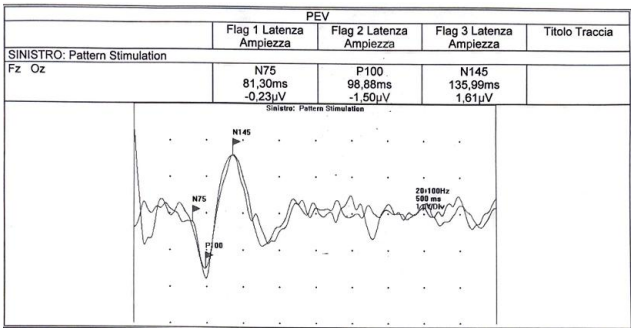
In this study, we report the case of a 59-year-old woman diagnosed with both CMT1B and NF2 who developed progressive asymmetrical bilateral visual loss (OD 8/10, OS 6/10) with mild language (semantic paraphasia) and memory deficits. The patient had undergone surgery for bilateral cataract 9 months earlier and had been regularly followed up for Irvine–Gass syndrome (IGS) in OS for several months. Despite the resolution of the edema, the patient did not regain 10/10 vision in OS and vision in the right eye (OD) actually progressively worsened as well. As part of a thorough workup to exclude causes associated with visual loss -including brain MRI and optical coherence tomography (OCT)- we performed an electroencephalogram (EEG) and visual evoked potentials (VEP). The evaluation also included neurobehavioral status examination, and a battery of neuropsychological tests (Activities of Daily Living, Instrumental activities of Daily living, MoCA Cognitive Assessment).

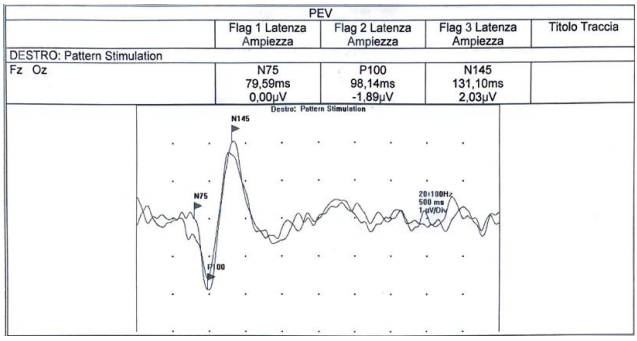
OUTCOME

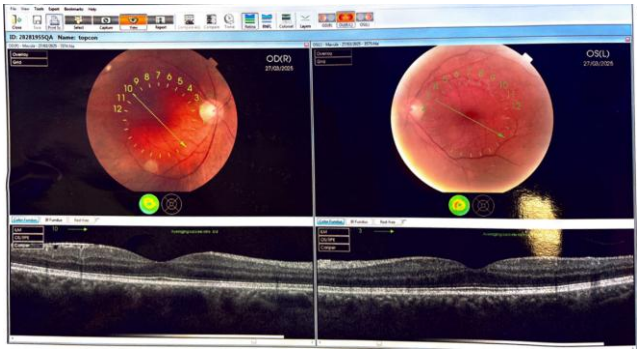
The patient's performance across neuropsychological tests did not reveal any cognitive impairment, despite brain MRI showing mild cortical atrophy, most evident in regions around the lateral sulcus. No intracranial space-occupying lesions, such as tumors, were found on MRI and OCT confirmed retinal and optic integrity. Regarding the electrophysiological examination, VEP testing was negative for abnormalities, thus excluding a visual pathway disorder. The EEG also didn't reveal anything relevant.

CONCLUSION

We report the first case of a patient with both CMT1B and NF2 who developed progressive cerebral visual impairment; this case is very uncommon. We suggest that CMT1B and NF2 mutations may play a pathophysiological role in promoting degeneration in the central nervous system: LZTR1 expression, which is mutated in NF2, is robust in the cortex, amygdala, hippocampus, and oligodendrocytes in the white matter. Further investigations such as an electroretinogram (ERG) and a PET tomography, could be helpful.







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