

Neurofibromatosis (NF1) and multiple sclerosis (MS) - a rare association

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Background: Neurofibromatosis 1 (NF1) is a genetic autosomal dominant neurocutaneous disorder. There are only a few cases of NF1 associated with multiple sclerosis (MS), 16.6% of the MS patients are carriers of the NF1 gene. The mutation in the oligodendrocyte myelin glycoprotein (OMG) gene is frequently associated with the primary progressive form of MS. Case description: Our patient is a 30 year female diagnosed with NF1, with a family history of neurofibromatosis (mother and grandmother), who presented cafe`-au-lait patches, Crown sign, Lisch nodules and dermal neurofibromas. In the last 13 years she developed 2 episodes of self limiting optic neuritis and a right cerebellar syndrome for which she was admitted in our clinic. The brain MRI showed multiple T2-weighted areas of altered signal in both cerebral hemispheres suggestive for MS and the spinal MRI –same type of lesions at the C2-C3 and C4-C5 level. Cerebrospinal fluid examination showed high levels of oligoclonal bands and IgG index. The visual-evoked potentials were significantly bilaterally delayed. Other autoimmune and infectious diseases were excluded and OMG gene was absent. Her symptoms ceased under corticosteroid therapy. She underwent immunomodulatory treatment for relapsing-remitting MS with no exacerbations (3 years now –EDSS=1.5p). Conclusions: The peculiarity of our case is the fact that NF1 usually goes along with primary progressive MS and no medullar lesions. Our patient has a relapsing-remitting form with a good response to immunomodulator treatment.