Very late-onset AQP4-positive NMO associated with severe autoimmune hemolytic anemia and thrombocytopenia: a case report

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Objective

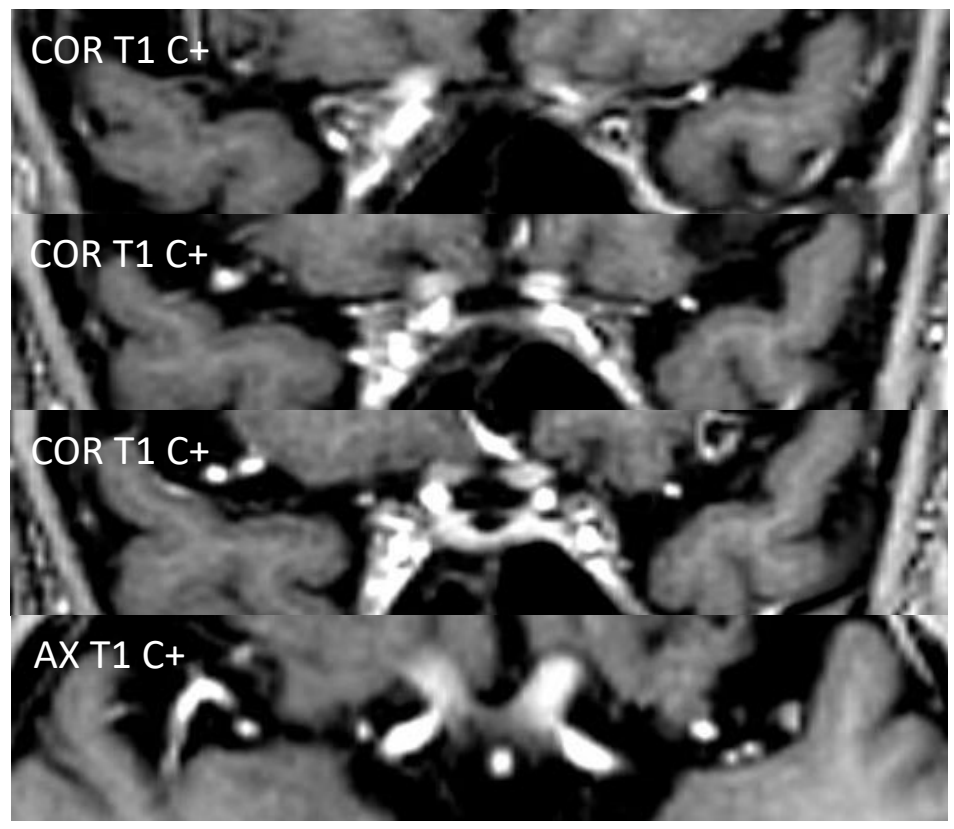
To report a rare case of very late-onset anti-AQP4 positive neuromyelitis optica spectrum disorder (NMOSD) associated with severe autoimmune hemolytic anemia (AIHA) and thrombocytopenia.

Materials

A 76-year-old man with a past history of chronic neuropsychiatric disorders and bladder cancer (4 years earlier) presented with subacute vision loss in the left eye, followed five days later by subacute vision loss in his right eye, rapidly progressing to complete bilateral amaurosis within one week, in absence of any pain on eyes movement. On admission, neurological examination was unremarkable, except for bilateral amaurosis and mydriasis.

Methods

Extensive blood tests (routine, direct/indirect Coombs test, deficiency screening, thyroid profile, infectious disease screening including Quantiferon TB gold [QTB], TPHA and VDRL, autoimmune screening including anti-AQP4 and anti-MOG antibodies in serum, tumor markers), peripheral blood smear, immunophenotyping, bone marrow aspiration, ophthalmological examination, brain MRI with contrast, CT total body with contrast, abdominal MRI with contrast, PET/CT total body with 18-FDG.



ophthalmological evaluation: VA 1/10 OD, hand movement OS, normal fundus; MRI: T2 hyperintensity, swelling and contrast enhancement of bilateral prechiasmatic optic nerves and chiasm (L>R); CT total body and abdominal MRI with contrast: enhancing pancreatic tail nodule; PET/CT total body with 18-FDG: negative; oncology consult ruled out malignancy and a paraneoplastic syndrome. A diagnosis of primary AQP4-IgG-positive NMOSD with associated autoimmune anemia and thrombocytopenia was made. The patient was treated with IV methylprednisolone (80 mg/day, 3 weeks), IVIG (0.6 g/kg/day, 3 days), and a prednisone taper, with a slow/progressive improvement of vision and hematological parameters (Hb 10.4 g/dL, PLT 146.000/µL) at discharge. IV Rituximab (1000 mg × 2) was scheduled.

Results

Tests showed: severe anemia (Hb 5.7 g/dL), thrombocytopenia (PLT 17.000/µL), positive direct/indirect Coombs tests; serum positivity for anti-AQP4 IgG, anti-TPO, ANA, ASMA, and IgM/IgG antiphospholipid antibodies; peripheral smear: rare atypical myeloid cells; immunophenotype: increased CD34+ cells; bone marrow aspirate: negative; QTB: positive (PCR and microscopy for BK on bronchial aspirate: negative); HBcAb: positive (HBsAg, HBsAb, and HBV-DNA negative); nonspecific elevation of CA19-9 and Chromogranin-A;

Discussion/Conclutions

This case highlights that, despite optic neuropathies in elderly patients are mostly ischemic or arteritic, an inflammatory anti-AQP4-associated etiology should be considered in absence of: vascular risk factors, clinical and laboratory features suggestive of arteritic anterior ischemic optic neuropathy (A-AION) and abnormal fundus findings.

References

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