Neuromyelitis Optica in Marfan Syndrome

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Neuromyelitis optica (NMO) is one of the differential diagnoses that should be considered in a patient with unilateral or bilateral loss of vision. It should be evaluated by history, examination, serological testing and neuroimaging studies.

We report a case of a 39-year-old gentleman who was known to have Marfan's syndrome and presented with progressive loss of vision in one eye followed by the other one within one month. Neurological examination showed bilateral optic neuritis (ON) with optic atrophy and unilateral upper motor neuron signs. CSF analysis was positive for NMO-IgG; MRI of the brain and spine showed enhancement in both optic nerves pathways and the optic chiasm with normal spine appearance.

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Neuromyelitis optica (NMO) is an uncommon severe autoimmune inflammatory demyelinating central nervous system disorder in which clinical spectrum ranges from severe attacks of recurrent isolated optic neuritis to extensive myelitis with bilateral neuritis. Visual impairment is common, blindness affecting at least one eye in 60-70% at a mean time of 5 years.

On the other hand, Marfan syndrome is an autosomal dominant systemic disorder of connective tissues. Individuals affected by the Marfan syndrome carry a mutation in one of their two copies of the gene that encodes the connective tissue protein fibrillin-1 (FBN1). Clinical diagnosis depends on a combination of major and minor signs defined in the revised 1996 Ghent criteria. The hallmark features are noted in the cardiovascular, skeletal and ocular systems.

The rarity of NMO with coexistence of genetic diseases is a challenge in diagnosis and therapy.

The prime objective is to identify any association or linkages between NMO and genetically inherited disorders, such as Marfan's syndrome and identify possible relationships that may influence diagnosis and treatment of both coexisting conditions in the future.

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