LEBERS HEREDITARY OPTIC NEUROPATHY "PLUS" SYNDROME MISDIAGNOSED AS MULTIPLE SCLEROSIS: A CASE REPORT

Safwan Zar MD, David Le BS, Azfar Niazi MD, Tariq Niazi MD, Antonio Quidgley-Nevares MD

Case Description:

51-year-old male with diagnosed *Lebers Hereditary Optic Neuropathy* presented to the acute care setting for ongoing lower extremity **weakness, numbness** and **functional decline**.

The patient was subsequently treated with intravenous **corticosteroids** for a suspected *Multiple Sclerosis (MS) flare up*.

Further follow up demonstrated a **further decline** in functional status and worsening symptoms. Patient was ruled out for *Neuromyelitis Optica (NMO) spectrum disorders*. Lumbar and thoracic MRI demonstrated abnormal T2 hyperintense signals along the dorsal columns, with contiguous enhancement of the lower cervical spine representing diffuse myelitis. In light of the **extraocular manifestations, MRI findings,** and **lack of improvement with corticosteroids** neurological consultation determined the diagnosis of *LHON-Plus Syndrome*. Patient subsequently underwent acute rehabilitation for functional decline.

LHON** with extraocular manifestations, **LHON plus**, is often misdiagnosed at **MS** or **NMO

Discussion:

LHON is caused by mitochondrial DNA mutations and has poor prognosis in regard to visual acuity. Very rarely, it is associated with **extraocular manifestations** and then referred to as *LHON-Plus Syndrome*. These patients are often misdiagnosed with *MS* or *NMO* due to the non-specified extra-ocular manifestations. These symptoms may include movement disorders, peripheral neuropathy, multiple sclerosis-like syndromes, brainstem and basal ganglia involvement, seizures and tremors. Our case highlights the course of a LHON patient whose extraocular presentation was consistent with the a rare LHON-Plus Syndrome originally misdiagnosed.

Conclusions:

This case is unique due to an atypical presentation of a rare *LHON-Plus Syndrome* with misdiagnosis of *MS*. There has been a reported link of misdiagnosis with *MS* given the similarities in signs and symptoms including **cord enhancements on imaging, ocular manifestations** and **neuropathic symptoms**. Other common misdiagnosis may include *NMO* spectrum disorders. Nonetheless, there is a need for heightened awareness of this rare form of *LHON* and its deceiving presentation. Faster recognition can lead to a timely diagnosis, initiation of proper clinical and pharmacological intervention along with prompt rehabilitation to optimize the patient's outcomes.