Marburg’s Disease and Neuromyelitis Optica in Brother/Sister
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Introduction

Marburg’s disease (acute MS)
• Rare variant of MS
• Monophasic, progressive course
• Multifocal neurological disturbances
• Rapid deterioration
• High mortality rate

Neuromyelitis optica (NMO)
• Distinct demyelinating entity
• Selective involvement of the optic nerves and spinal cord
• Rare involvement of the brain
• Extensive involvement of the spinal cord; more than 3 vertebral segments
• Progressive or relapsing course
• Immunosuppressive therapy

Methods:

Patient #1
38 year old man:
• No PMH
• Progressive right arm weakness
• One month later: LE weakness with rapid progression
• Four months later: wheelchair bound
• Brain MRI and LP: demyelination
• Eighteen months later: patient expired

Patient #2
Nine years later, patient’s younger sister at 43 y.o.:
• ATM treated with IV steroids and recovered
• In the following two years, two more episodes of ATM and one episode of left ON occurred
• MRI of the brain with and without gadolinium: unremarkable
• MRI of the spine: extensive demyelination at the spinal cord
• LP: no oligoclonal bands
• NMO-IgG: positive
• On immunosuppressive therapy
• Has moderate disability

Neuroimaging:

Fig 1. T2-weighted sequences, axial. Extensive white matter disease in the brain stem

Fig 2. T2-weighted sequences, sag. Signal changes in the thoracic cord.

Conclusions/Discussion:

Familial cases of multiple sclerosis between siblings or parents and children have been reported. A case of NMO in a mother and daughter and another case of NMO and MS between sisters have been documented. This is the first case report demonstrating the occurrence of two rare demyelinating diseases in a brother and sister. This rare occurrence suggests a complex genetic etiology and probable genetic heterogeneity even between members of the same family. It requires further testing and consideration.

Significance to MS:
Genetic risk in MS is low, but real. It calls for further investigation.

References:
Cabrera-Gomez JA, Ramon-Perez L. NMO and MS in sisters. Multiple Sclerosis 2008: October 15

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