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Anti-Neurofascin Antibody Associated Disease: A Case Series

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Mentor: Rana Zabad

Program: Neurological Sciences

Type: Original Research

Background: Autoantibodies against nodal and paranodal proteins, particularly anti-neurofascin (NF) antibodies, have been recently described in demyelinating disorders of the central and peripheral nervous system. There is an unmet need for further characterization of anti-NF associated demyelinating diseases. We describe the clinical and diagnostic findings of five patients with anti-NF antibodies, and attempt to identify features that might guide clinicians to check for them.

Methods: This is a retrospective chart review of five individuals seen at the University of Nebraska Medical Center Multiple Sclerosis Clinic who tested positive for serum anti-NF antibodies by western blot.

Results: In our series of five patients, the female to male ratio was 4:1, and the median age at presentation was 60 years (range: 56-70). Clinical presentations included incomplete transverse myelitis (n=2), progressive myelopathy (n=1), recurrent symmetric polyneuropathy (n=1), and non-specific neurological symptoms (n=1). Atypical features prompting further workup included co-existing upper and lower motor neuron features, older age at presentation with active disease, atypical spinal cord MRI

features and unusual CSF findings. Serum anti-NF antibody panel was positive for the NF-155 isoform in three patients (IgM n=2; IgG n=1), and the NF-140 isoform in two patients (IgG n=2).

Conclusions: Our case-series suggests that anti-NF antibodies are more commonly detected in the older populations and women. The clinical phenotype is pleiotropic. Atypical findings on MRI and paraclinical tests prompted further workup. Larger studies are needed to assess the impact of anti-NF antibodies on long-term clinical outcomes and their associated therapeutic implications. ■

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Seropositive Pediatric Autoimmune Encephalitis: A Single Center Experience

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Program: Neurology

Type: Original research

Background: The objective of the study is to describe the clinical characteristics and identify unique features associated with antibody-positive autoimmune encephalitis (AE).

Methods: Charts of children aged below 18-years diagnosed with AE between 2005 and 2020 were reviewed. Demographics, clinical characteristics, laboratory, imaging, electrophysiological findings and long-term-neurological-sequelae were collected.

Results: Among 53 AE cases, 19 had serum or CSF antibodies. Among antibody-positive

AE patients, there were 9 (47%) male and 10 (53%) female patients of whom; 10 (52.6%) had anti-NMDAR-antibodies, 5 (26%) anti-VGKC-antibodies and 4 (21%) demonstrated GAD65-antibodies. Clinical presentation included neuropsychiatric symptoms (68.4%), altered mental status (63%), movement disorders (57.9%), new-onset seizures (52.6%), speech difficulties (15.7%), status epilepticus (10.5%) and sleep dysfunction (5%). Two patients (10.5%) required ICU admission and 9 (47.3%) experienced relapses needing readmissions. MRI Brain was abnormal with T2 hyperintensities in 13 patients (31.5%) - 3 in parietal lobe, 2 in temporal lobe and 2 in frontal lobe. Seven patients (36.8%) had focal EEG findings. Nine patients (47.3%) had CSF pleocytosis and protein was normal in all patients. Acute management consisted of several different combination regimens including intravenous

steroids (89%), intravenous immunoglobulin (IVIG) (84%), rituximab (15.8%) and plasma exchange (PLEX) (10.5%). Maintenance regimen included IVIG (42%), Rituximab (36.8%) and 1 PLEX. Six patients (31.5%) attained complete remission and one patient underwent excision of ovarian teratoma. Long term sequelae included neurocognitive disturbances (52.6%), epilepsy (31.5%) and movement disorders (5%).

Conclusion: The most frequent presentation of AE in our cohort was neuropsychiatric disturbances, followed by altered mental status. Given diverse clinical presentations, one should maintain broad differential diagnosis when evaluating pediatric patients with subacute onset neurological symptoms. ■

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Risk Factors for Mortality Among Patients with Gout in the Veteran's Health Administration

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Program: Rheumatology

Type: Original Research

Background: A complete understanding of factors driving the increased all-cause mortality risk in patients with gout is lacking. The purpose of this study was to examine

risk factors associated with mortality among patients with gout in the Veteran's Health Administration (VHA).