Neuroimaging

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Understanding the influence of APOE-ε4 on magnetic resonance imaging-based hippocampal phenotypes in Alzheimer's disease and Lewy body dementia

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doi: 10.1017/cjn.2021.333

Background: The ε4-allele of apolipoprotein E (*APOE*-ε4) increases the risk not only for Alzheimer's disease (AD), but also for Parkinson's disease dementia and dementia with Lewy bodies (collectively, Lewy body dementia [LBD]). Hippocampal volume is an important neuroimaging biomarker for AD and LBD, although its association with *APOE*-ε4 is inconsistently reported. We investigated the association of *APOE*-ϵ4 with hippocampal atrophy quantified using magnetic resonance imaging in AD and LBD. **Methods:** Electronic databases (PubMed, Embase, PsycINFO, Scopus, Web of Science) were systematically searched for studies published up until December 31st, 2020. **Results:** Thirty-nine studies (25 cross-sectional, 14 longitudinal) were included. We observed that: (1) *APOE*-ϵ4 was associated with greater rate of hippocampal atrophy in AD and those who progressed from mild cognitive impairment to AD, (2) *APOE*-ϵ4 carriers showed greater involvement of cornu ammonis-1 hippocampal subfield versus non-carriers in AD, (3) *APOE*-ε4 may influence hippocampal atrophy in dementia with Lewy bodies, although longitudinal investigations are required, and (4) *APOE*-ϵ4 associated with earlier rather than very late expression of mediotemporal degeneration and memory-related neurocognitive impairment. **Conclusions:** The role of *APOE*-ε4 in modulating hippocampal phenotypes may be further clarified through more homogenous, well-powered, pathology-proven studies. Understanding the underlying mechanisms will facilitate development of prevention strategies targeting *APOE*-ε4.

Neuromuscular Disease and EMG

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A clinical mystery finally revealed

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doi: 10.1017/cjn.2021.334

Background: We report 2 brothers sharing FHL1 identified mutation. They presented in childhood with overlapping clinical features characterized by early onset stiffness and increased muscle definition with cardiac involvement. After 30 years of neurological followup, the diagnosis is finally revealed. **Methods:** At early ages, both had increased definition of upper trunk musculature. The older brother had hypophonic voice with raspy character, which is to our knowledge, not reported with this mutation before. He required a pacemaker for arrhythmias, while the younger developed congestive heart failure. **Results:** Their initial investigations failed to unveil a diagnosis, including a negative next generation sequencing (NGS) panel for AR LGMD. An expanded NGS sent on the older brother revealed he is hemizygous for 1770 bp deletion within FHL 1 gene, this deletion includes exon 7to 8, and confirmed on the other. **Conclusions:** First reported in 2008, FHL1 mutations result in phenotypically distinct neuromuscular disorders: X-linked myopathy with Postural Muscle Atrophy and generalized hypertrophy, X-linked dominant scapuloperoneal Myopathy, and Reducing Body Myopathy. Subsequently other phenotypes have been reported including Emery-Dreifuss muscular dystrophy and hypertrophic cardiomyopathy. Our patients present with a phenotype that had been reported with FHL1 mutation, highlighting the possible recognition of this presentation in aiding a diagnostic approach.

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Clinical and Electrophysiological characteristics of anti-nodal/paranodal antibodies in chronic inflammatory demyelinating polyradiculoneuropathy patients

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doi: 10.1017/cjn.2021.336

Background: CIDP is an autoimmune polyneuropathy. Antibodies against the Node of Ranvier have been described, NF155, NF140/186 and contactin-1. **Methods:** A retrospective review of patients with CIDP who tested positive for anti-nodal/paranodal antibodies via Western blot were evaluated. We have included 20 sero-negative CIDP patients. All patients met definite or probable EFNS criteria. clinical, electrophysiological data and response to treatment were obtained. **Results:** Forty-five patients tested positive for the antibodies. Sixteen were positive for NF155, 11 for NF140, 5 for CNTN1,11 were double positive for NF155 and NF140, and 3 were triple positive for NF155, NF140 and CNTN1.

Age of onset was similar in both seronegative (53.9 ± 3.1 yrs.) versus seropositive (52.3 ± 2.4 yrs.).

Chronic presentation manifested in 85% of seronegative, 80% of seropositive patients.Intrestingly,all triple-positive patients presented with a more acute presentation (i.e,<8 wks.)

7/20 seronegative (35%),1/16 NF155, 6/11 NF140,1/5 contactin, 2/11 of double-positive, 3/3 of triple-positive (28%,13/46) responded to IVIg. **Conclusions:** No major clinical or electrophysiological differences between groups. triple-positive patients showed 100% response to IVIg.These results cast doubt on the specificity of the Western blot as a clinico-electrophysiologic discriminator. Future testing with cell-based assays will likely provide a robust measure that will guide treatment decision.