Effect of ApoE isoforms on mitochondria in Alzheimer disease

APOE ε4 and mitochondrial dysfunction are risk factors for late-onset Alzheimer disease. In a postmortem study of the brain, APOE ε4 was associated with impaired mitochondrial structure and function, oxidative stress, and synaptic integrity. Mitochondrial proteins and synaptic protein were strongly associated with cognitive performance.

From editorialists Qin et al.: “These data provide the most compelling evidence to date that directly links ApoE4 to mitochondrial alterations in the human brain and confirm the importance of mitochondrial homeostasis, thereby providing clues for future mechanistic studies.”

The CLN3 Disease Staging System: A new tool for clinical research in Batten disease

Natural history data from 108 participants in an 18-year-long study were used to develop the CLN3 Disease Staging System to categorize disease stages in patients with juvenile neuronal ceroid lipofuscinoses. This staging system has potential utility for staging clinical research.

Homozygous nonsense variant in LRIF1 associated with facioscapulohumeral muscular dystrophy

The authors identified a patient with facioscapulohumeral muscular dystrophy (FSHD) and biallelic truncating variants in LRIF1, demonstrating that LRIF1 is required for DUX4 repression in skeletal muscle, and reinforcing the current hypothesis that FSHD is caused by D4Z4 chromatin relaxation resulting in inappropriate DUX4 expression.

From editorialists Johnson and Ankala: “While it is appropriate that early clinical developments focus on the most common form of FSHD, FSHD1, we would encourage investigators to expand as rapidly as possible to these other causes of FSHD.”
Plasma neurofilament light levels are associated with risk of disability in multiple sclerosis

Plasma neurofilament light chain (NfL), a marker of axonal damage, was measured in 4,385 patients during the early stages of multiple sclerosis (MS), and was found to be predictive of future Expanded Disability Status Scale score worsening. Plasma NfL can serve as a reliable biomarker to assess risk of long-term disability worsening in MS.

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NB: “Pearls & Oy-sters: Contrast-induced encephalopathy following coronary angiography: A rare stroke mimic,” p. e2491. To check out other Resident & Fellow Section Pearls & Oy-sters articles, point your browser to Neurology.org/N and click on the link to the Resident & Fellow Section. At the end of the issue, check out the NeuroImage illustrating intramural patchy gadolinium enhancement in the subcortical and leptomeningeal arteries and veins of a patient with cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. This week also includes a Humanities in Neurology piece titled “Space traveler.”

Exosomal neurofilament light: A prognostic biomarker for remote symptoms after mild traumatic brain injury? (see p. 1016)

1. Exosomal neurofilament light: A prognostic biomarker for remote symptoms after mild traumatic brain injury?
2. What’s Trending: Managing migraine, part 2

In the first segment, Dr. David Lapides talks with Dr. Kimbra Kenney about her paper on exosomal neurofilament light as a biomarker following traumatic brain injury. In the second part of the podcast, Dr. Jason Crowell continues his talk with Dr. David Watson on managing migraine.

Disclosures can be found at Neurology.org.