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Research

Pharmacotherapy for Pain in Inherited Erythromelalgia 659
Geha and colleagues determine whether pain in inherited erythromelalgia can be attenuated via pharmacotherapy guided by genomic analysis and functional profiling. The main outcomes and measures were behavioral assessment of pain, functional magnetic resonance imaging, and assessment of firing in dorsal root ganglion neurons carrying S241T mutant channels. Their results demonstrate that pharmacotherapy guided by genomic analysis, molecular modeling, and functional profiling can attenuate neuropathic pain in patients carrying the S241T mutation. Editorial perspective is provided by Juan M. Pascual, MD, PhD.

Extrapyramidal Movement Disorders in Mitochondrial Disease 668
Martikainen and coauthors describe the phenotype, genetic etiology, and investigation of extrapyramidal movement disorders in a large and well-defined mitochondrial disease cohort. They report that dystonia, often associated with Leigh syndrome, was the most common extrapyramidal movement disorder among pediatric patients with mitochondrial disease and that parkinsonism was the most prevalent extrapyramidal movement disorder in adults and was commonly associated with POLG mutations. Editorial perspective is provided by Robert D. S. Pitceathly, MD, PhD.

Cerebellar Atrophy and Anti-NMDAR Encephalitis 706
Iizuka et al report the long-term clinical implications of diffuse cerebral atrophy and cerebellar atrophy in anti-N-methyl-D-aspartate receptor (NMDAR) encephalitis. A retrospective observational study and long-term imaging investigation were conducted. Fifteen patients with anti-NMDAR encephalitis admitted to Kitasato University Hospital between January 1, 1999, and December 31, 2014, were included. Editorial perspective is provided by Maarten J. Titulaer, MD, PhD.

Nonfluent/Agrammatic Primary Progressive Aphasia Variants 733
Santos-Santos and colleagues characterize the neurological, cognitive, and neuroimaging features of patients with nonfluent/agrammatic primary progressive aphasia (nfvPPA)—in whom either progressive supranuclear palsy (PSP) or corticobasal degeneration (CBD) was eventually confirmed at autopsy. Inclusion criteria for the study were a clinical diagnosis of nfvPPA, the availability of speech, language, and cognitive testing for at least 1 evaluation; magnetic resonance imaging within 6 months of initial evaluation; and a postmortem pathological diagnosis of PSP or CBD. In patients presenting with nfvPPA, they found that the presence of early severe dysarthria, relatively selective white matter atrophy at presentation, and a greater rate of change in the brainstem measured by longitudinal imaging may be useful for differentiating underlying PSP from CBD pathology during life.