In Focus

Spotlight on the July 27 Issue

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Familial neuromyelitis optica

This paper describes a case series involving the demographic, clinical, neuroimaging, and NMO-IgG serologic status of 12 multiplex neuromyelitis optica (NMO) pedigrees in a total of 25 affected individuals. Familial NMO was indistinguishable from sporadic NMO based on clinical symptoms, age at onset, sex distribution, and frequency of NMO-IgG detection, suggesting complex genetic susceptibility in NMO.

See p. 310

Redefining dysferlinopathy phenotypes based on clinical findings and muscle imaging studies

This retrospective study assessed lower limb MRI in 29 patients to identify the pattern of muscle impairment and quantify involvement. The finding that proximal and distal muscles were already impaired at onset in both Miyoshi myopathy and limb-girdle muscular

dystrophy 2B favors grouping all phenotypes under the term *dysferlin myopathy*.

See p. 316; Editorial, p. 298

Quality of life declines after first ischemic stroke: The Northern Manhattan Study

The long-term quality of life of stroke patients is not well characterized. In this population-based study of 525 patients with first ischemic stroke, there was a linear decline in quality of life over 5 years independent of recurrent vascular events and common predictors.

See p. 328

Use of antiepileptic drugs in epilepsy and the risk of self-harm or suicidal behavior

This observational study investigated the risk of suicidal behavior associated with the use of antiepileptic drugs in 44,300 patients with epilepsy. The authors found an increased risk in some of the newer antiepileptic drugs, but not from the most commonly used antiepileptic drugs.

See p. 335; Editorial, p. 300

Physical activities and future risk of Parkinson disease

Physical activity was evaluated in 213,701 participants in the NIH-AARP Diet and Health. The authors found that higher levels of moderate to vigorous physical activities were associated with lower risk of Parkinson disease; further analysis suggests that this finding might not be due to reverse causality.

See p. 341

Cortical neuroanatomic correlates of symptom severity in primary progressive aphasia

Forty right-handed patients with primary progressive aphasia had performance-based language testing using the Progressive Aphasia Severity Scale (PASS) and MRI scans. These data support the value of PASS and MRI as

novel clinical measures and as markers for potential use in clinical research and trials in primary progressive aphasia.

See p. 358

Nongenetic factors influence severity of episodic ataxia type 1 in monozygotic twins

The authors evaluated 3 families with episodic ataxia type 1 phenotype, 2 of which included monozygotic twins. They sequenced the *KCNA1* gene and studied the biophysical consequences of the mutations in HEK cells. The occurrence of epilepsy in 1 of 2 families with the F414S mutation suggests interplay of *KCNA1* with other genetic factors.

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