July 13 Highlights

Genetics
Hedera et al. (p. 44) describe the phenotype of hereditary spastic paraplegia (HSP) linked to chromosome 8q23-24. Other than increased severity, no clinical features distinguished the family they describe from those with autosomal dominant HSP linked to other chromosomes. The authors question whether the multiple genes responsible for these clinically indistinguishable disorders involve a common biochemical cascade. Mitochondrial disturbance, postulated to be a common factor of HSP, is not a feature of the new gene. Martínez-Murillo et al. (p. 50) characterize a new and apparently common gene locus for recessive familial spastic paraparesis linked to chromosome 15q. In their accompanying editorial, Figlewicz and Bird (p. 5) review the genetics of HSP and highlight the critical issues that must be settled. ◆ Three articles and the editorial by Tournier-Lasserve (p. 3) relate to CACNA1A mutations. Jen et al. (p. 34) describe a nonsense mutation in the gene that causes episodic ataxia and weakness as well as episodic hemiplegia with or without migraine. Battistini et al. (p. 38) performed lineage analysis of the CACNA1A gene in two siblings who experienced typical hemiplegic migraine attacks, sometimes associated with altered consciousness and fever. Both individuals also experienced a permanent, late onset, cerebellar ataxia with cerebellar atrophy. Acetazolamide effectively treated the episodic symptoms but had no effect on the progressive ataxia. The authors speculate that the combination of episodic and permanent deficits could depend on the variety of functions of calcium channels and their distribution in the nervous system. Carrera et al. (p. 26) studied Italian families with familial hemiplegic migraine. They detected several new genetic variants and noted that the new missense mutation G4644T is associated with milder symptoms. ◆ By demonstrating the loss of heterozygosity for the Von Hippel–Lindau (VHL) gene in a VHL patient's endolymphatic sac tumor, Kawahara et al. (p. 208) establish the tumor's relationship to this disease and define the mechanism of tumorigenesis. In his accompanying editorial, Roach (p. 7) reviews theories of tumorigenesis in VHL and speculates as to what role inactivation of the somatic VHL gene may have in patients without VHL who, nonetheless, develop similar tumors.

Stroke
Adams et al. (p. 126) compared the baseline NIH Stroke Scale (NIHSS) score and the stroke subtype used in the Trial of Org 10172 in Acute Stroke Treatment (TOAST) as predictors after ischemic stroke. They conclude that the NIHSS score strongly predicts the likelihood of a patient's recovery. Only the subtype of lacunar stroke predicts outcomes independent of the NIHSS score. The same research team (p. 122) performed an exploratory analysis of outcomes at 7 days and 3 months among patients enrolled in the TOAST study who had an ischemic stroke in the hemisphere ipsilateral to an occlusion or a stenosis of greater than 50% of the ICA, identified by carotid duplex imaging. They conclude that early identification by duplex imaging might improve selection of patients for treatment with emergent anticoagulation.

Movement disorders
Moro et al. (p. 85) describe seven parkinsonian patients who underwent bilateral high frequency subthalamic nucleus stimulation. The procedure was well-tolerated and efficacious. ◆ Blanchet et al. (p. 91) performed a double-blind, placebo-controlled, two-arm crossover study of high-dose transdermal beta-estradiol in eight postmenopausal women with mild to moderate PD. The treatment had an antiparkinsonian effect without consistently altering dyskinesias.

Neuromuscular/ neuroimmunology
Leppert et al. (p. 62) studied the expression of matrix metalloproteinases (MMPs) in chronic inflammatory demyelinating polyneuropathy (CIDP) and nonsystemic vasculitic neuropathy (NSVN). They conclude that the upregulation of MMP-2 and -9 is a specific feature of these conditions and suggest that selective inhibitors of these enzymes could prevent tissue damage. The similar increases in these MMPs in both types of neuropathies argues against a primary role of MMPs in demyelination. ◆ Kieseier et al. (p. 20) review evidence pointing to MMPs as new targets for treatment of inflammatory demyelination. ◆ Hadden et al. (p. 57) report the results of a randomized trial of interferon beta-1a in CIDP in which the agent was safe but not efficacious in treatment-resistant CIDP.

Epilepsy
Leidy et al. (p. 162) compare the health-related quality of life (HRQOL) of a nonsurgical sample of adults with epilepsy to that of age- and gender-matched normal subjects. Seizure-free adults can have HRQOL levels comparable to those of the general population. As seizure frequency increases, patients report more impaired HRQOL, regardless of other factors.

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