In Focus

Spotlight on the March 12 Issue

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The challenge of juvenile Huntington disease: To test or not to test

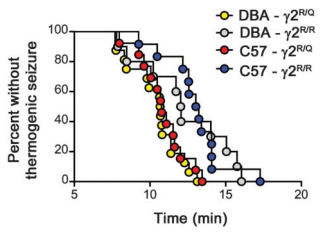
Genetically testing a child at risk of Huntington disease (HD) can be challenging. The authors analyzed 76 patients with suspected juvenile HD, concluding that a family history of HD rather than the nature of presenting symptoms should primarily guide the decision. Nevertheless, a proportion of symptomatic children with positive family history will test negative.

See p. 990

From editorialists Lehman & Nance: "The greatest challenge to the clinician is the 10- to 20-year-old patient presenting with behavioral symptoms or cognitive changes, as attentional difficulties, depression, and anxiety are common disorders in the general population."

See p. 976

Multiple molecular mechanisms for a single $GABA_A$ mutation in epilepsy



A $GABA_A$ receptor mutation causes two distinct seizure phenotypes, a common scenario in genetic epilepsy. The authors used mouse models to demonstrate that two independent molecular mechanisms were operative, providing a novel explanation for clinical heterogeneity. Building such conceptual frameworks brings the promise of personalized medicine closer.

See p. 1003

$CHADS_2$, CHA_2DS_2-VASc, and long-term stroke outcome in patients without atrial fibrillation

$CHADS_2$ and CHA_2DS_2-VASc assess stroke risk in patients with atrial fibrillation; this study extends the scores' role to predict stroke outcome in stroke patients without atrial fibrillation. Among 1,756 stroke patients, 5-year mortality was higher in the high-risk $CHADS_2$ and CHA_2DS_2-VASc subgroups.

See p. 1009

Disability in multiple sclerosis: A reference for patients and clinicians

A Disability Expectancy Table, based on ~28,000 patient records from the North American Research Committee on Multiple Sclerosis, affords a detailed overview of disability outcomes in multiple sclerosis during the first 45 years of disease. This allows clinicians and patients to determine how an individual's disability compares to others with similar disease duration.

See p. 1018

Cognitive impairment in MS: Impact of white matter integrity, gray matter volume, and lesions

Conventional MRI and diffusion tensor imaging data were acquired from 55 MS patients (35 cognitively preserved, 20 cognitively impaired) and 30 controls. In cognitively impaired patients, white matter damage, measured with diffusion tensor imaging, was more extensive in areas important for cognition (e.g., thalamus).

See p. 1025

Modulation of neural activation following treatment of hepatic encephalopathy

Twenty-two patients with biopsy-proven cirrhosis of differing etiology and previous minimal hepatic encephalopathy were treated with oral L-ornithine L-aspartate for 4 weeks. Baseline and 4-week clinical review, blood chemistry, and psychometric evaluation (Psychometric Hepatic Encephalopathy Score and Cognitive Drug Research Score) were performed in addition to functional imaging. Default mode network activity can be modified by hepatic encephalopathy therapy.

See p. 1041

Prediction of dementia in MCI patients based on core diagnostic markers for Alzheimer disease

The 3 core biomarkers were collected and measured with standardized procedures. When patients with mild cognitive impairment (MCI) underwent clinical follow-up, 29 progressed to dementia, while 44 remained stable. Memory clinics able to collect and measure medial temporal atrophy, temporoparietal hypometabolism, and CSF biomarkers can give accurate diagnostic answers to worried patients with MCI.

See p. 1048; Editorial, p. 978

NB: "Ethical and quality pitfalls in electronic health records," see p. 1057. To check out other Contemporary Issues, point your browser to www.neurology.org.

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