



In Focus

Spotlight on the March 13 Issue

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SMN1 gene duplications are associated with sporadic ALS

The authors conducted a genetic investigation in 847 patients with amyotrophic lateral sclerosis (ALS) and 984 controls, using multiplexed ligation-dependent probe amplification assays, examining the effects on disease susceptibility and course. These data provide evidence for a role of common *SMN1* duplications in ALS and raise new questions regarding the disease mechanisms involved.

See p. 776

From editorialist Basil T. Darras: "Whether or not SMN overexpression occurs in vivo in human motor neurons via SMN1 duplications remains uncertain, but taken together, these recently described associations could stimulate research endeavors. . . ."

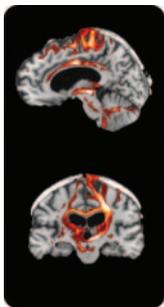
See p. 770

The LRRK2 G2019S mutation is associated with Parkinson disease and concomitant non-skin cancers

Among 490 patients with Parkinson disease (PD) genotyped for the *LRRK2* G2019S mutation, non-skin cancer frequency was double in carriers vs noncarriers, especially among Ashkenazi Jews. These findings suggest that *LRRK2* carriers should be offered screening for non-skin cancers.

See p. 781; Editorial, p. 772

The tremor network targeted by successful VIM deep brain stimulation in humans



Twelve patients undergoing ventral intermediate (VIM) nucleus stimulation for debilitating tremor were tested using noninvasive diffusion tractography from tremor-suppressive VIM electrode contacts. These patients provided a unique opportunity to assess an electrophysiologically defined seed region in human thalamus, a technique that is

usually restricted to animal research.

See p. 787

Early EEG correlates of neuronal injury after brain anoxia

In 61 adults treated with therapeutic hypothermia (TH) after cardiac arrest, multichannel EEG recorded during TH was assessed for background reactivity and continuity, presence of epileptiform transients, and correlated with serum neuron-specific enolase collected at 24–48 hours after cardiac arrest. Early EEG after cardiac arrest reflects the extent of neuronal damage.

See p. 796; Editorial, p. 774

Homozygous c.14576G>A variant of RNF213 predicts early-onset and severe form of moyamoya disease

A comprehensive analysis of *RNF213* genotype and clinical symptoms was undertaken in 204 Japanese persons with moyamoya disease. Homozygotes of c.14576GA variant had earlier onset with more frequent occurrence of infarctions at initial presentation and posterior cerebral artery involvement. The homozygous c.14576GA variant in *RNF213* may be a good DNA biomarker for predicting severe moyamoya disease.

See p. 803

Predictors of hemorrhage volume and disability after perimesencephalic subarachnoid hemorrhage

The authors identified 31 patients with subarachnoid hemorrhage (SAH) without an identifiable aneurysm; platelet activity was measured on admission and aspirin use was recorded. Discontinuous venous drainage and reduced platelet activity were associated with increased SAH volume and hydrocephalus. These factors may explain large SAH and reduce the need for repeated invasive imaging in such patients.

See p. 811

Relationship between etiology and covert cognition in the minimally conscious state

Using an EEG task, 30% of 23 minimally conscious patients consistently produced appropriate responses to motor imagery commands. When separated according to etiology, none of the 8 nontraumatic patients were able to produce such signs of command-following, showing that external behaviors of patients in the minimally conscious state were not necessarily an accurate reflection of their true cognitive capacities.

See p. 816

NB: "The evidence for a role of B cells in multiple sclerosis" (see p. 823). To check out other Views & Reviews, point your browser to www.neurology.org.

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