In Focus

Spotlight on the July 17 Issue

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GBA-associated PD: Neurodegeneration, altered membrane metabolism, and lack of energy failure

MR spectroscopic imaging was performed in 13 patients with Parkinson disease (PD) with heterozygous *GBA* mutations and in 19 controls to investigate metabolite concentrations within the mesostriatal regions of PD pathology. The neurodegenerative process in GBA-PD was associated with alterations of membrane phospholipid metabolism and with abnormal α -synuclein aggregation.

See p. 213

From editorialists Albin & Dauer: "Looking ahead, defining PS subtypes and refining clinical trial methods to allow evaluation of subtype-specific effects may be necessary to make progress in developing disease-modifying therapies."

See p. 202

Novel late-onset Alzheimer disease loci variants associate with brain gene expression

This study assessed the influence of genetic variants on brain levels of genes at novel LOAD GWAS loci, identifying significant genetic associations for brain levels of LOAD risk genes, *CLU*, *MS4A4A*, and *ABCA7*. These results suggest that changes in brain levels may be an important disease mechanism for some of the novel LOAD risk genes.

See p. 221; Editorial, p. 204

Low PiB PET retention in presence of pathologic CSF biomarkers in Arctic APP mutation carriers

The authors examined 7 APP_{Arctic} mutation carriers, 7 controls, and 7 patients with sporadic Alzheimer disease (AD). The lack of fibrillar β -amyloid ($A\beta$) as visualized by PiB PET in APP_{Arctic} mutation carriers suggests that other forms of $A\beta$, such as oligomers and protofibrils, are important for the pathologic processes leading to clinical AD.

See p. 229; Editorial, p. 206

Three-day CSF drainage barely reduces ventricular size in normal pressure hydrocephalus

Fifteen patients were investigated with 1.5-T MRI before and after external lumbar drainage (ELD). Ventricular volume was measured. Clinical improvement occurred in patients with normal pressure hydrocephalus after ELD despite unaltered ventricles, suggesting that ventricular size is of little relevance for postshunt improvement or determining shunt function.

See p. 237

Polymorphisms in the glial glutamate transporter SLC1A2 are associated with essential tremor

Using the Affymetrix Genome-Wide SNP Array 6.0, the authors conducted a 2-stage study in 964 cases and 1,539 controls to identify genetic variants associated with essential tremor (ET). The glial glutamate reuptake transporter *SLC1A2* was identified as a possible susceptibility gene, suggesting glutamatergic overexcitation in the pathogenesis of ET.

See p. 243

Networks involved in seizure initiation: A reading epilepsy case studied with EEG-fMRI and MEG

Mechanisms causing seizures are different from interictal epileptiform activity and are difficult to study. The authors reported MEG and EEG-fMRI data from a patient with reading epilepsy that supports the concept of a unique site in the piriform cortex modulating seizure activity within a network of cortical and subcortical regions.

See p. 249

Interferon- β and serum 25-hydroxyvitamin D interact to modulate relapse risk in MS

Serum 25-hydroxyvitamin D [25(OH)D] was measured biannually in 178 persons with clinically definite multiple sclerosis. Interferon- β was associated with reduced risk of relapse, and this effect may be modified by a positive effect of interferon- β on serum 25(OH)D levels.

See p. 254; Editorial, p. 208; see also pp. 261 and 267

Vitamin D and disease activity in multiple sclerosis before and during interferon- β treatment

This study followed 88 patients with relapsing-remitting multiple sclerosis (MS) for 6 months. In untreated MS patients, increased levels of 25-hydroxyvitamin D were associated with radiologic disease activity irrespective of their HLA-DRB1*15 status.

See p. 267; Editorial, p. 208; see also pp. 254 and 261

NB: "Trick or treat? Showing patients with functional (psychogenic) motor symptoms their physical signs," see p. 282. To check out other *Contemporary Issues in Neurologic Practice*, point your browser to www.neurology.org.

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