In Focus

Spotlight on the April 23 Issue

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Incidence of atrial fibrillation detected by implantable loop recorders in unexplained stroke

Ischemic stroke etiology is frequently unclear despite thorough investigation, although atrial fibrillation (AF) is difficult to exclude. Using implantable loop recorders (ILRs) that can detect AF (Reveal XT), AF was identified in 25% of unexplained stroke cases. ILRs may have a central role in the investigation of patients with unexplained stroke.

See p. 1546; Editorial, p. 1542

Higher serum glucose levels are associated with cerebral hypometabolism in Alzheimer regions

This study investigated 124 cognitively normal persons with a first-degree family history of Alzheimer disease (AD), including 61 APOE ϵ 4 noncarriers and 63 carriers. Findings suggest that the risk of higher serum glucose levels may be independent of APOE ϵ 4 status, raising additional questions about the role of the metabolic process in the predisposition to AD.

See p. 1557

Pregnancy outcomes following gabapentin use: Results of a prospective comparative cohort study

The authors compared neonatal outcomes from >220 infants exposed to gabapentin during pregnancy with an equal number of unexposed infants. It is evident that pregnant women worldwide are taking gabapentin, and this evidence showing that gabapentin does not increase the risk of major malformations may assist in calculating the benefits of using this drug during pregnancy.

See p. 1565

ADORA2A polymorphism predisposes children to encephalopathy with febrile status epilepticus

The authors analyzed 4 adenosine A2A receptor (ADORA2A) single nucleotide polymorphisms in 85 patients with acute encephalopathy with biphasic seizures and late reduced diffusion (AESD). Homozygous haplotype A diplotype of ADORA2A was associated with AESD and may alter the intracellular adenosine/cyclic adenosine monophosphate cascade, promoting seizures and excitotoxic brain damage.

See p. 1571

Cognitive changes predict functional decline in ALS: A population-based longitudinal study

This study enrolled 186 ALS patients with no evidence of C9orf72 hexanucleotide repeat expansion and conducted follow-up neuropsychological assessment in 144 patients; controls were also studied. Non-C9orf72-associated ALS was characterized by nonoverlapping cognitive subgroups with different disease trajectories, with important implications for models of ALS pathogenesis, relevant for future clinical trials.

See p. 1590

Rare cell capture technology for the diagnosis of leptomeningeal metastasis in solid tumors

CSF samples from 60 patients were analyzed, with the main cohort consisting of 51 patients with solid tumors; all had lumbar puncture for clinical suspicion of leptomeningeal metastasis (LM). Rare cell capture technology is an accurate, novel method for the detection of LM in solid tumors, potentially providing earlier

diagnostic confirmation.

See p. 1598; Comment, p. 1603

The p.L302P mutation in the lysosomal enzyme gene SMPD1 is a risk factor for Parkinson disease

The genetic background of Parkinson disease (PD) is complex, involving multiple genes. Two cohorts of Ashkenazi Jewish patients from Tel Aviv and New York were analyzed for founder mutations that cause lysosomal storage disorders. The Niemann-Pick SMPD1 p.L302P mutation conferred a 9.4-fold increased risk for Parkinson disease, emphasizing the importance of the lysosome for PD pathogenesis.

See p. 1606

From editorialist Nutan Sharma: "The finding that a specific mutation in the SMPD1 enzyme is associated with an increased risk of PD gives further support to the hypothesis that defects in the ALP [autophagy-lysosomal pathway] play a role in the pathogenesis of PD..."

See p. 1544

Hyperdopaminergic crises in familial dysautonomia: A randomized trial of carbidopa

The authors examined 12 patients with familial dysautonomia to assess the safety of carbidopa and evaluate its antiemetic efficacy. Carbidopa at an average daily dose of 480 mg (range 325 to 600 mg/day) was well tolerated and effective in reducing nausea and retching in patients with familial dysautonomia.

See p. 1611

NB: "Changing practice: Residents' adoption of the atraumatic lumbar puncture needle," see p. e180. To check out other Resident & Fellow Education Research submissions, point your browser to www.neurology.org and click on the link to the Residents & Fellow Section.

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