Everolimus alters white matter diffusion in tuberous sclerosis complex

This trial of everolimus demonstrated significant reductions in subependymal giant cell astrocytoma volume and decreased seizure frequency. Of 28 subjects, 20 had sufficient diffusion tensor imaging data. Fractional anisotropy and radial diffusivity changes after treatment with everolimus suggests that the genetic defect of tuberous sclerosis complex may be modified pharmacologically.

From editorialists Ess & Roach: “Targeting of tuber-less regions in tuberous sclerosis complex may seem paradoxical, but it may ultimately open the door to more effective therapies.”

Antigen microarrays identify CNS-produced autoantibodies in RRMS

The authors used antigen arrays to analyze the reactivity of antibodies in matched serum and CSF samples of 20 patients with untreated relapsing-remitting multiple sclerosis (RRMS), 26 patients with methylprednisolone-treated RRMS, and 20 controls. Their data provides a new avenue to investigate antibody responses in the CNS, which may help monitor disease progression and response to therapy.

Solitary sclerosis: Progressive myelopathy from solitary demyelinating lesion

Seven patients developed progressive myelopathy, 4 with typical CSF findings of multiple sclerosis, attributable to a single CNS lesion typically at the cervicomедullary junction. Multiple sclerosis may be the correct diagnosis for progressive myelopathy even when “dissemination in space” is not fulfilled.

A randomized trial of varenicline (Chantix) for the treatment of spinocerebellar ataxia type 3

Twenty patients with genetically confirmed SCA3 were randomized to receive either varenicline or placebo over a period of 8 weeks. This study showed that varenicline was tolerated well and improved axial symptoms and rapid alternating movements in patients with SCA3 as measured by SARA subscores.

GLUT1 mutations are a rare cause of familial idiopathic generalized epilepsy

Glucose transporter type 1 (GLUT1) mutations were identified in 9 family members affected by absence epilepsy from early childhood to adulthood. Functional assays predicted reduced glucose transport across the blood-brain barrier. GLUT1 mutations should be considered in difficult-to-treat absence seizures, with the ketogenic diet considered for treatment.

Chitinase enzyme activity in CSF is a powerful biomarker of Alzheimer disease

The authors analyzed the level of chitinase activity, N-acetylglucosaminidase activity, stathmin, and EF-1/H9251 in CSF of 94 patients with AD, 41 patients with non-AD dementia, and 40 controls. Chitinase activity in CSF exhibited a sensitivity of 91.5% and specificity of 72.5% for AD, which was superior to the diagnostic power of β-amyloid and tau.

VIEWS & REVIEWS

Discussions about treatment restrictions in chronic neurologic diseases: A structured review

The authors performed a literature search for empirical studies about discussions and decisions to restrict treatment in amyotrophic lateral sclerosis, primary malignant brain tumors, multiple sclerosis, stroke, Parkinson disease, and Alzheimer disease. The currently available empirical data suggest that discussions were mainly triggered by life-threatening situations.

NB: “Hyperammonemic encephalopathy: Time course of MRI diffusion changes,” see p. 600. To check out other NeuroImages, point your browser to www.neurology.org.