In Focus

Spotlight on the October 15 Issue

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Editor-in-Chief, Neurology®

Deletions in *GRID2* lead to a recessive syndrome of cerebellar ataxia and tonic upgaze in humans

The authors performed genetic analyses and Sanger sequencing, identifying a homozygous deletion of *GRID2* exon 4 in the index family and compound heterozygous deletions involving *GRID2* exon 2 in a second family. Biallelic deletions of *GRID2* may lead to a syndrome of cerebellar ataxia and tonic upgaze in affected individuals.

See p. 1378

Prevalence of white matter lesions and stroke in children with migraine

Of 926 patients diagnosed with migraine, 375 had MRIs and 115 had abnormalities, with 39 having white matter lesions. The prevalence of white matter lesions is more common in pediatric migraine with aura, but they appear to be benign and are not associated with stroke.

See p. 1387

Epstein-Barr virus in oral shedding of children with multiple sclerosis

The authors used PCR-based assays to detect viral DNA in the monthly mouth swabs of 22 pediatric patients with multiple sclerosis (MS) and 77 controls. Children with MS demonstrated abnormally increased rates of Epstein-Barr virus (EBV) viral reactivation and a broader range of genetic variants, suggesting a selective impairment in their immunologic control of EBV.

See p. 1392

Natalizumab affects the T-cell receptor repertoire in patients with multiple sclerosis

T-cell receptor (TCR) repertoire in blood and CSF was assessed in 59 patients with multiple sclerosis (MS) treated with natalizumab, 5 cases of natalizumab-associated progressive multifocal leukoencephalopathy (PML), 17 patients with MS not treated with natalizumab, and 12 controls. TCR repertoire restrictions in CSF of patients treated with natalizumab reflected an altered immune surveillance of the CNS, contributing to an increased risk of developing PML.

See p. 1400

From editorialists Hohlfeld & Stüve: "... the observation that some but not all PML-related clonal expansions occurred in both the blood and CSF compartment supports the view that the anti-JCV immune response has a systemic and sequestered component."

See p. 1372

Preclinical AD predicts decline in memory and executive functions in subjective complaints

In 132 patients with subjective complaints, those with CSF evidence of preclinical AD at baseline showed decline in memory, executive functions, and global cognition. Slopes in stages 1 and 2 were similar, while baseline memory performance was worse in stage 2 only. Clinical progression increased when patients reached a later preclinical AD stage.

See p. 1409

Limbic hyperconnectivity in the vegetative state

The authors analyzed MRI data from 11 patients in a vegetative state and 7 patients in a minimally conscious state along with controls. There was increased limbic connectivity that was more complex than previously thought, which may lead to new treatment strategies to restore brain functions in these conditions.

See p. 1417

Duplications of 17q12 can cause familial fever-related epilepsy syndromes

The authors analyzed 433 patients with a broad range of epilepsy phenotypes, identifying 2 families in which a 17q12 duplication segregated with fever-related epilepsy. An inherited copy number variation in these self-limiting fever-sensitive epilepsy syndromes suggests a novel pathomechanism involved in familial febrile seizures and generalized epilepsy with febrile seizures plus.

See p. 1434

NB: "Brainstem PML lesion mimicking MS plaque in a natalizumab-treated MS patient," see p. 1470. To check out other NeuroImages, point your browser to www.neurology.org.

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