Dopamine transporter SPECT in fragile X premutation carriers

Ceravolo et al. studied four fragile X premutation carriers with parkinsonism using SPECT with ^{123}I-FP-CIT. There was evidence of preserved presynaptic nigrostriatal function.

see page 1971

Cerebral microbleeds in TIA and ischemic stroke

Cerebral microbleeds, detected by gradient-echo MRI, may be associated with fragile, bleeding-prone small vessels. Werring et al. found microbleeds in 24% of patients with ischemic stroke, but in only 2% of patients with TIA.

see page 1914

Classification of malformations of cortical development

Barkovich et al. propose a revised classification of malformations of cortical development. This revision updates and expands the classification using genotype as the basis for classification when the genotype-phenotype is adequately understood. A section on genetic testing is also included.

see page 1873

Idiopathic acute transverse myelitis (ATM)

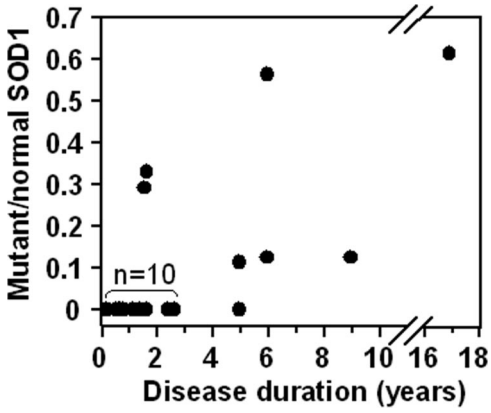
De Seze et al. studied 45 patients with idiopathic acute transverse myelopathy (ATM) in a group of 288 patients with ATM (15.6%). These patients formed a homogeneous group in terms of clinical and MRI data but the prognosis was highly variable.

see page 1950

The editorial by Cree and Wingerchuk notes that the clinical requirements for idiopathic ATM include bilateral sensory, motor, or autonomic dysfunction referable to spinal cord, with a defined sensory level that progresses to a nadir over 4 to 21 days from onset. Evidence of an inflammatory etiology is also required: gadolinium-DPTA enhancement within the cord, CSF pleocytosis, or IgG index elevation. Because these criteria are based on expert opinion, validation is needed. De Seze et al. have demonstrated that these criteria identify a relatively homogenous patient cohort and may define a subset of patients who share a common pathogenesis. However, MS and NMO cases comprised nearly 30% of this retrospective cohort. Moreover, the boundary between ATM secondary to a rheumatologic disorder and limited versions of NMO with coexistent nonspecific autoantibodies, such as anti-SSA, are not well defined.

see page 1857

Instability of mutant SOD1 and rapid FALS progression



Using liquid chromatography electrospray ionization mass spectrometry, Sato et al. analyzed mutant SOD1 proteins in erythrocytes from 29 patients with 22 different mutations, and found that turnover of mutant SOD1 correlated with a shorter disease survival time.

see page 1954

The editorial by Koji Yamanaka and Don W. Cleveland notes that mutations in superoxide dismutase (SOD1) cause 20% of familial ALS cases. Some mutations are fully active, and others completely inactive with no correlation between retention of dismutase activity and age at disease onset or duration of disease. Moreover, expression of ALS-linked SOD1 mutations in rodents provokes progressive motor neuron disease independent of dismutase activity, while total absence of SOD1 by gene deletion does not cause motor neuron disease. Sato et al. report what may be a pivotal, and counterintuitive, discovery concerning a toxic property shared among many mutants. An accelerated disease course was found for mutants that are less stable. That patients accumulating less mutant have more rapid disease course implies that it is the misfolded, unstable form(s) of SOD1 mutants that contribute to toxicity. However, despite its apparent importance for progression, SOD1 mutant stability is not correlated with disease onset. An essential extension of the current work will be to test the stability of the SOD1 mutants in the tissues most affected in ALS.

see page 1859

Multiple sclerosis (MS) and pregnancy, delivery, and birth outcome

Dahl et al. studied the effect of maternal MS on pregnancy, delivery, and birth outcome. Mothers with MS had a higher proportion of neonates small for gestational age, and also frequent induction and operative interventions during delivery.

see page 1961

New guidelines for diagnosis and management of DLB

The International Consortium on Dementia with Lewy bodies (McKeith et al.) has revised criteria for its clinical and pathologic diagnosis taking account of both Lewy- and Alzheimer-related pathologies. Additional clinical features and investigative findings suggestive of DLB are introduced. The limited data on patient management are reviewed.

see page 1863

Spheroid body myopathy caused by a mutation in TTID

Spheroid body myopathy (SBM) is a rare, autosomal dominant disorder. A mutation in the titin immunoglobulin domain protein (TTID) was detected in all 19 affected members of a single kindred. Mutations in TTID also cause limb-girdle muscular dystrophy 1A and are associated with myofibrillar myopathy. Foroud et al. propose that SBM is a myotilinopathy.

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