Mental retardation with cerebellar hypoplasia

Zanni et al. analyzed the oligophrenin 1 gene (*OPHN1*) in male patients with mental retardation with (17 patients) or without (196 patients) known cerebellar anomalies. The authors identified four novel OPHN1 mutations associated with distinguishing clinical and neuroimaging features. Of particular interest is the presence of cerebellar vermis and hemisphere hypoplasia.

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The editorial by Higgins and Topaloglu notes that besides raising issues whether individuals with OPHN1 gene mutations have syndromic-X-linked mental retardation, Zanni et al. identify a problem that plagues neurologists and neurogeneticists in the clinic. What is the correct diagnostic approach when presented with a child with X-linked mental retardation? Obtaining an MRI of the brain is a good start but if a cerebellar anomaly exists, the algorithm becomes complicated. A detailed knowledge of neuroimaging, genetics, and molecular neurobiology will raise many questions but not as many as how to decide whether the patient has a syndrome. A good beginning is to arrive at the clinicoradiologic diagnosis of X-linked oligophrenic vermian dysgenesis by identifying X-linked transmission of mental retardation in the family pedigree and analyzing the MRI for specific cerebellar anomalies.

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Seizure control in young children taking oxcarbazepine

Piña-Garza et al. report that high-dose oxcarbazepine (60 mg/kg/day) reduced seizure frequency more effectively than low-dose oxcarbazepine (10 mg/kg/day) in very young children who were already taking up to two concomitant antiepileptic drugs.

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The editorial by Shinnar and Pellock notes that in partial seizures in older children one can usually presume that drugs effective in adults with partial seizures will also be effective. In neonates and infants, no such assumptions can be made. There are few drugs and in particular few AEDs that have obtained a pediatric indication, particularly in infants and younger children—despite financial incentives to pharmaceutical manufacturers. Once a drug is approved for use in adults, there are major barriers to performing trials in children. While providing important data on the value of high-dose oxcarbazepine there are difficulties in performing such studies: over 30 centers were needed in a relatively common childhood epilepsy. Moreover, the study was not blinded, thus introducing potential bias in the assessment of adverse events.

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Genetic variation of *Campylobacter:* Guillain-Barré vs Fisher syndromes

Koga et al. report that a polymorphism of a *Campylobacter jejuni* gene, *cst-II*, is associated with clinical features of Guillain-Barré syndrome. The *cst-II* (Asn51) strains carried a GQ1b epitope and were prone to cause Fisher syndrome associated with anti-GQ1b IgG antibody.

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The editorial by Wokke and van den Berg considers the molecular mimicry hypothesis of the pathogenesis of the Guillain-Barré syndrome (GBS): the immune system of the patient is deceived by bacterial ganglioside-like epitopes that mimic peripheral nerve components and triggers inflammatory responses against peripheral nerve. Cross-reactive anti-ganglioside antibodies may be directed against myelin, Schwann cell components, and axonal membranes. Koga et al. provide important evidence that genetic polymorphisms of antecedent infectious agents determine the production of specific autoantibodies and clinical manifestations in a post-infectious disorder.

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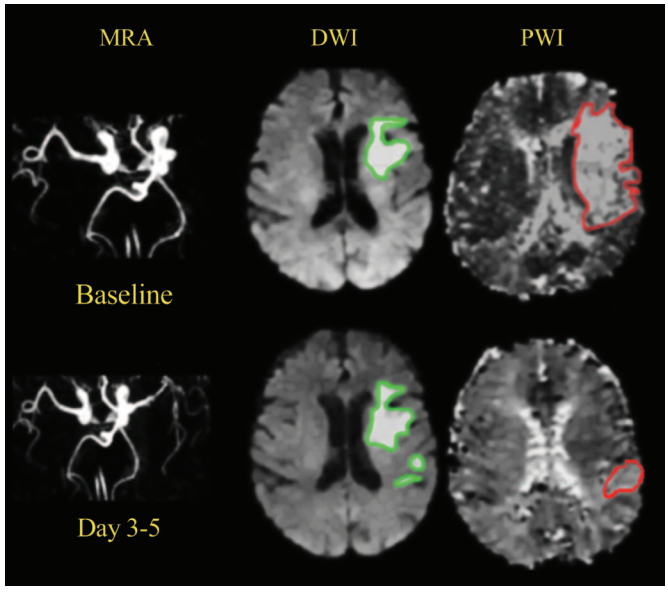
Hematocrit and tissue fate in acute stroke



Using serial diffusion- and perfusion-weighted MRI in 64 patients with acute stroke, Allport et al. demonstrated that an elevated hematocrit was associated with reduced odds of reperfusion and greater infarct size. This effect was independent of smoking, glucose, baseline diffusion and perfusion lesion volumes, and rtPA administration.

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Statin use and cognitive decline in the elderly

Studying an elderly community cohort, Bernick et al. found that statin drug use was associated with a slight reduction in cognitive decline vs nonusers over a 7-year period. The only MRI variables that differentiated statin users were a lower number of accumulated silent infarcts.

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Surrogate consent for high risk research?

Research consent for incompetent subjects lacks explicit policy guidance in most jurisdictions. Kim et al. surveyed persons at elevated risk for Alzheimer disease and found that the majority support surrogate consent for research even when the risk and burdens to subjects are substantial.

see page 1395

Patent foramen ovale in migraine with aura

In a cross-sectional case control study, Schwerzmann et al. studied the characteristics of cardiac right-to-left cardiac shunts in patients with migraine with aura using transesophageal echocardiography. Nearly half of the patients with migraine with aura had a patent foramen ovale, and shunt size larger than in controls without migraine with aura.

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Botulinum toxin treatment for cervical dystonia

A controlled study by Comella et al. directly compared two serotypes of botulinum toxin for the treatment of cervical dystonia. They found similar efficacy for serotypes A and B, with a modest prolongation of benefit using serotype A.

see page 1423

Daytime sleepiness and PD

In the Honolulu-Asia Aging Study Abbott et al. examined the relationship between excessive daytime sleepiness (EDS) and the development of Parkinson disease (PD). EDS was assessed in 3,078 elderly men from 1991 to 1993. Men were followed for incident PD to 2001. Findings suggest that EDS can predate PD.

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Ictal monoparesis with primary somatosensory lesion

Matsumoto et al. report three patients with ictal monoparesis of the arm. A well-circumscribed lesion in the primary arm somatosensory area was associated with epileptic activity.

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POLG mutations in Alpers syndrome

Nguyen et al. identified mutations in the mitochondrial DNA polymerase (*POLG*) as the cause of Alpers syndrome. Here they summarize nine causative mutations in *POLG*. Genetic testing may now be possible for this neurodegenerative disease of children and young adults.

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Long-term 24-hour duodenal levodopa infusion

Nyholm et al. treated five patients with PD with continuous 24-hour duodenal levodopa infusion for 13 to 37 months. Motor responses were stable and without increased side effects. Daily dosage increased slightly (14%) over the study period.

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Levodopa addiction in PD

Levodopa may be addictive. Borek et al. describe two parkinsonian patients, one of whom was quadriplegic, for whom psychological benefit rather than motor response was seen with levodopa.

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