In Focus

Spotlight on the July 10 Issue

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Exome sequencing as a diagnostic tool in a case of undiagnosed juvenile-onset GM₁-gangliosidosis

The authors used exome sequencing and SNP arrays to identify a rare neurogenetic disorder in a child and emphasized that the use of exome sequencing for neurogenetic cases eluding diagnosis was more appropriate than continued prolonged and costly single-gene analyses.

See p. 123; Editorial, p. 112; see also pp. 127 and 192

Exome sequencing in an SCA14 family demonstrates its utility in diagnosing heterogeneous diseases

This paper demonstrates the cost-effectiveness of exome sequencing to screen rapidly genetically heterogeneous disorders such as the inherited ataxias. The pure ataxia family investigated had a large number of candidate genes that were rapidly screened; a novel SCA14 mutation was identified.

See p. 127; Editorial, p. 112; see also pp. 123 and 192

Simulation-based education with mastery learning improves residents' lumbar puncture skills

Fifty-eight internal medicine residents with limited clinical experience completed lumbar puncture (LP) simulation-based education. These residents were compared to 36 clinically experienced neurology residents on an LP skills examination. Neurology residents scored lower than simulator-trained residents, with fewer neurology residents competent to perform a simulated LP.

See p. 132; Editorial, p. 115

Lower use of carotid artery imaging at minority-serving hospitals

This study obtained data from chart reviews of veterans hospitalized with ischemic stroke. Hospitals serving a greater proportion of minorities were more likely to exhibit racial disparities in obtaining carotid artery imaging. Because disparities were more pronounced at specific sites, local solutions are needed to identify and address local barriers.

See p. 138

From editorialists Benson & Freeman: "... we may someday overcome persistent disparities in health that go far beyond race alone, thereby improving the health of all citizens."

See p. 117

Falls and fractures in patients chronically treated with antiepileptic drugs

This study highlights a higher fracture risk in 150 epilepsy outpatients on antiepileptic medications, compared to 506 controls, with the highest risk among chronic users. Female users fell more frequently than female controls. Patients with epilepsy should be assessed for history of falls and fracture so that appropriate management strategies can be instituted.

See p. 145; Editorial, p. 119

Do acute phase markers explain body temperature and brain temperature after ischemic stroke?

The authors investigated the relationship between circulating markers of inflammation with brain and body temperature after stroke. Higher brain temperature in 44 acute stroke patients was associated with higher levels of circulating inflammatory markers, though the relationship with body temperature was complex.

See p. 152

Abnormal whole-brain functional networks in homogeneous acute mild traumatic brain injury

Thirty-five patients with acute mild traumatic brain injury (MTBI) and 35 controls underwent resting-state fMRI,

susceptibility weighted imaging, neuropsychological testing, and postconcussive symptom assessments. Whole-brain functional connectivity was altered within 4 weeks after MTBI, suggesting that changes in functional networks underlie the cognitive deficits and postconcussive complaints reported by patients with MTBI.

See p. 175

NB: Resident & Fellow Media and Book Reviews: *The Human Brain in 1492 Pieces: Structure, Vasculature and Tracts*, see p. e10. To check out other Resident & Fellow submissions, point your browser to www.neurology.org and click on the link to the Resident & Fellow Section.

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