

even if no specific diagnosis is suspected. Unique statistical characteristics of LRs allow for a wide application in different clinical settings.

EFFECT OF DURATION OF STATUS CONVULSION ON NEURONAL APOPTOSIS AND EARLY APOPTOTIC EVENTS IN HIPPOCAMPUS OF RATS

Submitted by Li Jiang

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OBJECTIVE: Our goal was to explore the influence of duration of status epilepticus on neuronal apoptosis, mitochondrial membrane potential, and cytochrome c release in hippocampus in Wistar rats after status epilepticus (SE).

METHODS: SE that lasted for 30 minutes or 3 hours was induced by intraperitoneal injection with lithium chloride and pilocarpine. Rats were killed at different time points. The apoptosis, mitochondrial membrane potential, and intracellular cytochrome c level were investigated by flow cytometry.

RESULTS: The proportion of apoptotic cells, the decrease of mitochondrial membrane potential, and the release of intracellular cytochrome c significantly changed 30 minutes after 30-minute SE. The peak level was at the 12th hour after SE and 6th hour after SE in apoptosis and the 2 early apoptotic events, respectively. Compared with the same time point after 30-minute SE, the levels of apoptosis and the 2 early apoptotic events after 3-hour SE were much higher. The neuronal apoptosis and the 2 early apoptotic events in hippocampus after SE had a positive correlation with the duration of SE in partial correlation analysis.

CONCLUSIONS: Severe seizures could induce the changes of neuronal apoptosis and the early apoptotic events in hippocampus after SE; the longer the duration of SE, the more serious the change of apoptosis and early apoptotic events were.

PROTEOLIPID PROTEIN 1 GENE MUTATION IN CHINESE PATIENTS WITH PELIZAEUS-MERZBACHER DISEASE

Submitted by Yuwu Jiang

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INTRODUCTION: Pelizaeus-Merzbacher disease (PMD) is a rare X-linked recessive disorder that presents with nystagmus, impaired motor development, ataxia, and progressive spasticity.

OBJECTIVE: The objective of this study was to analyze the proteolipid protein 1 (*PLP1*) gene in 6 Chinese patients with PMD.

METHODS: Six unrelated Chinese patients had PMD (P1–P6), and 14 individuals were from family P2. Of these 6 patients, 3 had transitional, 2 had classical, and 1 had connate PMD according to the clinical and MRI features. Genomic DNA was extracted from peripheral blood samples. Gene dosage was determined by multiplex ligation-dependent probe amplification. All 7 exons and exon-intron boundaries of *PLP1* gene were amplified and analyzed by direct DNA sequencing.

RESULTS: *PLP1* duplications were identified in patients 1 through 4 with PMD. Their mothers were *PLP1* duplication carriers. Both duplication carriers and normal genotypes of *PLP1* were identified in the family members of patient P2. A c.517C → T (p. P173S) hemizygous missense mutation in exon 4 was found in patient 5, and his mother was a heterozygote of this mutation.

CONCLUSIONS: We identified 4 gene duplications and 1 missense mutation (p. P173S) of *PLP1* gene in 5 Chinese patients with PMD. This is the first report about *PLP1* mutations in patients with PMD in China.

IRON-DEFICIENCY ANEMIA IS ASSOCIATED WITH CEREBRAL SINOVENOUS THROMBOSIS: A CASE SERIES

Submitted by Fotini D. Kavadas

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INTRODUCTION: Iron-deficiency anemia is a relatively common but preventable condition in children that may have significant adverse implications on children's growth and development. Iron-deficiency anemia has also been sporadically reported in association with cerebral sinovenous thrombosis (CSVT).

OBJECTIVE: The objective of this study was to describe the largest case series to date of iron-deficiency anemia in association with CSVT as an advocacy measure for its prevention in children.

METHODS: Patients were identified through the Canadian Pediatric Ischemic Stroke Registry database (Toronto site). Included were patients who were older than 1 month to 18 years, met criteria for iron-deficiency anemia, and had radiographically confirmed CSVT.

**PROTEOLIPID PROTEIN 1 GENE MUTATION IN CHINESE PATIENTS
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Pediatrics 2008;121;S146

DOI: 10.1542/peds.2007-2022ZZZZZ

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