Clinical commentary

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A *de novo GABRB2* variant associated with myoclonic status epilepticus and rhythmic high-amplitude delta with superimposed (poly) spikes (RHADS)

Aiko Nishikawa [1], Yui Otani [1], Susumu Ito [1], Satoru Nagata [1], Mutsuki Shiota [2], Jun-ichi Takanashi [3], Mitsuko Nakashima [4], Hirotomo Saitsu [4], Naomichi Matsumoto [5], Hirokazu Oguni [1]

[1] Department of Pediatrics, Tokyo Women's Medical University, Tokyo, Japan
[2] Department of Pediatric Neurology, Fuji-Toranomon seikeigeka hospital, Shizuoka, Japan
[3] Department of Pediatrics, Tokyo Women's Medical University, Yachiyo Medical Center, Chiba, Japan
[4] Biochemistry Department, Hamamatsu University School of Medicine, Hamamatsu, Japan
[5] Department of Human Genetics, Yokohama City University Graduate School of Medicine, Yokohama, Japan

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Epileptic Disorders

- Alpers syndrome is an early-onset neurodegenerative disorder with a poor prognosis, characterized by the triad of developmental regression, intractable epilepsy, and hepatic dysfunction.
- Treatment-resistant epileptic seizures develop in the form of focal, multifocal or myoclonic seizures evolving into epilepsia partialis continua or myoclonic status epilepticus.
- Rhythmic high-amplitude delta with superimposed (poly) spikes (RHADS) on EEG is specific for this syndrome.
- As the use of VPA accelerates fatal hepatic failure, an early diagnosis is mandatory.

- GABA-A receptors play a cardinal role in controlling neuronal excitability in the central nervous system.
- Mutations in genes encoding α1、β2、β3 and γ2 subunits have been found in patients with epileptic encephalopathies.
- GABA-A β2 missense variants can cause, not only early myoclonic encephalopathy, but also an Alpers syndrome phenotype characterized by treatment-resistant myoclonic status epilepticus and RHADS.

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