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SEIZURE DISORDERS

PANAYIOTOPOULOS SYNDROME VS. GERDS

Investigators at Sapienza University of Rome, Italy, describe 12 children with Panayiotopoulos syndrome (PS) misdiagnosed as gastroesophageal reflux disease (GERD). They were referred to the gastroenterological service at the mean age of 6 years (from 4 to 10 years) because of drug-resistant, recurrent nausea, retching, and vomiting. Attacks occurred mainly during sleep; 3 children also had episodes of severe headache resembling migraine with visual aura (spots, zigzag lines, colored lights, and blobs). When seen by a pediatric neurologist because of persistence of symptoms, despite antacids, antireflux diet, and sleep position, an interictal EEG showed brief multifocal and generalized sharp and slow-wave paroxysms in all patients. Brain MRI was normal. In 5 patients treated with antiepileptic drugs, episodes of vomiting, headache, and EEG abnormalities were controlled. (Parisi P, Pacchiarotti C, Ferretti A, et al. Gastroesophageal reflux disease vs. Panayiotopoulos syndrome: an underestimated misdiagnosis in pediatric age? **Epilepsy Behav** 2014 Sep 27;41C:6-10).

COMMENTARY. Panayiotopoulos syndrome (PS) is defined as a benign age-related focal seizure disorder occurring in early and mid-childhood. It is characterized by seizures, often prolonged with predominantly autonomic symptoms, and by an EEG that shows shifting and/or multiple foci, often with occipital predominance [1]. Prognosis is usually favorable, but rarely, severe bradycardia and cardiac arrest have been reported [2]. In addition to GERD, nonepileptic disorders misdiagnosed as PS include syncope, and migraine. Cyclic vomiting is described as an epilepsy in an early report [3], but is usually considered an atypical migraine or periodic syndrome.

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AED treatment of PS and rolandic epilepsy (RE). A clinical practice survey in the UK among 590 pediatricians who treat epilepsy found that 40% of 132 respondents reported non-treatment of PS and RE because of low frequency of seizures and parent/child preferences. They estimated 233 new cases of PS and 751 new RE cases, annually. Carbamazepine is the preferred older, and levetiracetam the preferred newer AED in randomized controlled trials [4].

References.
1. Berg AT, et al. Epilepsia. 2010 Apr;51(4):676-85.
2. Moseley B, et al. Epilepsy Behav. 2013 Mar;26(3):375-85.
3. Millichap JG, Lombroso CT, Lennox WG. Pediatrics. 1955 Jun;15(6):705-14.
4. Mellish LC, et al. Arch Dis Child. 2014 Sep 8. [Epub ahead of print].

KCNB1 MUTATIONS IN EPILEPTIC ENCEPHALOPATHY

Researchers at Scripps Research Institute, San Diego, and other centers in California; Johns Hopkins, Baltimore; Vanderbilt University, TN; and Northwestern University Feinberg School of Medicine, Chicago, IL; searched for de novo mutations in a family with a sporadic case of epileptic encephalopathy. The cause was determined using whole exome sequencing (WES) and whole genome sequencing (WGS). A de novo missense mutation in KCNB1 was identified that encodes the K2.1 voltage-gated potassium channel. Subsequently, 2 additional patients were identified with epileptic encephalopathy and de novo KCNB1 missense mutations that cause a similar pattern of K2.1 dysfunction. Clinical WES may be useful for diagnosis of epileptic encephalopathies of unknown etiology. (Torkamani A, Bersell K, et al. De novo KCNB1 mutations in epileptic encephalopathy. **Ann Neurol** 2014 Oct;76(4):529-40).

COMMENTARY. Researchers at University of Arizona, Tucson, previously explored the utility of WES and identified causal de novo variants in genes of 7 of 10 children with sporadic epilepsy, refractory seizures, developmental delay, or epileptic encephalopathy. These probands all presented with seizures within the first 6 months of life, and 6 have intractable seizures. The genes affected included SCN1A, CDKL5, EEF1A2, and KCNH5 [1]. The present finding of de novo KCNB1 mutations as a cause of K2.1 dysfunction expands the locus heterogeneity associated with epileptic encephalopathies [2].

References.
1. Veeramah KR, et al. Epilepsia. 2013 Jul;54(7):1270-81.
2. Torkamani A, et al. Ann Neurol. 2014 Oct;76(4):529-40.

TRANSITION CARE TO ADULT EPILEPSY CENTERS

Epileptologists at University of Toronto and University of Saskatchewan, Canada, evaluated the complexity of epilepsy patients transitioned from child to adult care between tertiary centers compared to patients transferred from the community. Patients aged from 18 to 25 years were divided into 2 groups: Group 1 comprised 170 patients referred from the pediatric tertiary center; and Group 2 had 132 patients referred from the