

COMMENT. A Canadian study considering strategies for transitioning to adult care for youth with Lennox-Gastaut syndrome and related disorders (Camfield PR, et al. **Epilepsia** 2011 Aug;52 Suppl 5:21-7) found that an adult practitioner took less time with the patient and family, and the adult provider was not familiar with the medical disorder. A survey of 133 symposium attendees indicates much dissatisfaction with the process of transition, especially for patients with intellectual handicap. Suggestions to improve transition include identifying a willing adult service, a multidisciplinary approach, adolescent clinics, and attention to vocational training and/or special education.

## **MOVEMENT DISORDERS**

### **HYPEREKPLEXIA, APNEAS, DEVELOPMENTAL DELAY, AND GENETIC CORRELATIONS**

Investigators at Swansea University and other centers in the UK, Australia, and Belgium studied the genotype-phenotype correlations in 97 individuals with a clinical diagnosis of hyperekplexia; 61 cases had mutations in GLRA1, 24 cases in SLC6A5 and 12 in GLRB. All gene-positive cases presented in the neonatal period and clonazepam was effective treatment in 95%. Hyperekplexia is a predominantly recessive inheritance, and is dominant in 16%. In 35 gene-negative cases, presentation was after the first month of life.

The characteristic symptoms of hyperekplexia are ‘stiffness, startles and stumbles.’ In addition, 50 of 89 patients had apnea attacks and 47 of 92 were developmentally delayed. Recurrent infantile apneas occurred more frequently in patients with SLC6A5 mutations than in those with GLRA1 mutations. Developmental delay occurred more frequently in patients with GLRB and SLC6A5 mutations than in those with GLRA1 mutations; 92% of GLRB cases had a mild to severe delay in speech acquisition. The developmental delay especially in speech may represent failure of developmental neural networks or migration defects. (Thomas RH, Chung S-K, Wood SE, et al. Genotype-phenotype correlations in hyperekplexia: apnoeas, learning difficulties and speech delay. **Brain** 2013 Oct;136(Pt 10):3085-95). (Response: Dr Rhys H Thomas, Institute of Life Science, College of Medicine, Swansea University, Singleton Park, Swansea, SA2 8PP. E-mail: Rhys.Thomas@swansea.ac.uk).

COMMENT. Hyperekplexia was first described by Kok O, and Bruyn GW (**Lancet** 1962;279(7243):1359) in 29 members of one family and occurred as a dominant autosomal transmission. Hypertonia is present at birth and becomes less pronounced during the first year of life but later leads to repeated falls. The name “hereditary stiff baby syndrome” was used by Lingam S, et al. (**Am J Dis Child** 1981 Oct;135(10):909-11). The child has a fixed stare and an expression of anxiety. Hypertonia diminishes during sleep and increases with the slightest psychic or tactile stimulus. Nose tapping elicits the hyperekplexic response and is included in the neonatal exam of infants at risk. Attacks of hypertonia that involve the respiratory muscles can lead to apneas that endanger life. The EMG shows persistent activity abolished by diazepam or clonazepam; the EEG is normal.

Early genetic testing is recommended for symptomatic hyperekplexic neonates and possibly preconception counseling for those at risk for GLRB and SLC6A5 mutations. Recessive inheritance of hyperekplexia is associated with an increased risk of learning difficulties and developmental delay, particularly in speech acquisition. Severe recurrent neonatal apneas occur in approximately 50% of cases, particularly those with mutations in GLRB and SLC6A5. Stiffness, startle and stumble are the cardinal features of hyperekplexia.

## **DEMYELINATING DISORDERS**

### **SINGLE CENTER EXPERIENCE OF ACUTE DISSEMINATED ENCEPHALOMYELITIS**

Investigators at Department of Pediatrics, Neurology Division, Adana Medical Research Center; and Division of Child Neurology, Ankara, Turkey, retrospectively evaluated 15 children with acute disseminated encephalomyelitis (ADEM) in children from the center in Adana. ADEM was seasonal, 73.3% cases presenting in winter or spring. The majority (13/15, 86.7%) had an acute febrile upper respiratory illness 2 to 40 days before presentation. Five children had serological evidence of specific triggers: mycoplasma (2), influenza-A (H1N1) (1 child), and Epstein-Barr virus (2 children). One patient had received a combined vaccine (DTPP-Haemophilus influenzae B) 6 weeks before onset. Gait disturbance (12/15, 80%) was the most common presenting symptom, followed by altered consciousness (10/15, 66.7%), fever (7/15, 46.7%), headache (4/15, 26.7%), seizures (4/15, 26.7%), meningismus (4/15, 26.7%), and vomiting (3/15, 20%). EEG recorded in 8 patients showed generalized slowing in 3 patients and focal epileptiform discharges in 1. CT scan obtained in 14 patients showed lesions in only 3 cases, whereas MRI revealed cerebral lesions in all 15 patients (with complete resolution following treatment in 12, partial in 2). Treatment in all cases was a standard protocol of 3 to 5 days of IV methylprednisolone and IV immunoglobulin for patients with persistent deterioration. Oseltamivir and clarithromycin were administered in patients with influenza-A and mycoplasma. Follow-up evaluation ranged from 0.6 to 4 years (median 1.8 years). Neurologic symptoms and signs resolved in 13 patients; one patient had severe neurologic sequelae, and one had recurrent attacks and a final diagnosis of MS. (Erol I, Ozkale Y, Alkan O, Alehan F. Acute disseminated encephalomyelitis in children and adolescents: a single center experience. **Pediatr Neurol** 2013 Oct;49(4):266-73). (Response: Dr Erol, Division of Pediatric Neurology, Adana Teaching and Medical Research Center, Baskent University Faculty of Medicine, Adana, Turkey. E-mail: ilknur\_erol@yahoo.com).

COMMENT. ADEM is an inflammatory demyelinating disease of the CNS that follows an infection or vaccination in three-quarters of the cases.

**Post-vaccination ADEM.** Several vaccines have been implicated including rabies, DTP, smallpox, measles, mumps, rubella, pertussis, and influenza (Huynh W et al. Post-vaccination encephalomyelitis: literature review and illustrative case. **J Clin Neurosci** 2008 Dec;15(12):1315-22). A patient presenting with bilateral optic