EARLY ONSET WEST SYNDROME WITH HYPOMYELINATION, COLOBOMA AND *SPTAN1* MUTATION

Investigators from Ljubljana, Slovenia report an 8-month-old female infant with hypotonia, lack of visual attention, early onset epileptic encephalopathy, and severe developmental delay. She presented with myoclonic jerks at 6 weeks and infantile spasms at age 3.5 months, accompanied by a hypsarrhythmia pattern EEG. Seizures were resistant to pyridoxine, levetiracetam, vigabatrin, and hydrocortisone. Topiramate was of some benefit, but the EEG progressed to a suppression-burst-like pattern. Brain MRI at 10 weeks of age revealed hypomyelination in the internal capsule and central regions, thinning of corpus callosum, and cortical atrophy. Eye exam showed dysplastic, coloboma-like optic discs. A heterozygous deletion was detected in *SPTAN1* gene. (Writzl K, Primec ZR, Strazisar BG, et al. Early onset West syndrome with severe hypomyelination and coloboma-like optic discs in a girl with *SPTAN1* mutation. **Epilepsia** 2012 June;53(6):e106-e110). (Respond: Karin Writzl MD, PhD, Institute of Medical Genetics, University Medical Centre, 1000 Ljubljana, Slovenia. E-mail: karinwritzl@gmail.com).

COMMENT. Mutations in the alpha-II-spectrin (*SPTANI*) gene are previously described in 2 Japanese children with early onset West syndrome, hypomyelination, and hypotonia. (Saitsu et al, 2010, cited in Slovenia article). The Slovenia case expands the phenotypic spectrum to a Caucasian child and with coloboma-like optic discs. All 3 patients had deceleration of rate of head growth, resulting in microcephaly. Patients with infantile spasms show mutations to several genes, and the present report supports the theory of a symptomatic basis for the majority of West syndrome cases. (Paciorkowski AR et al. Genetic and biological classification of infantile spasms. **Pediatr Neurol** 2011 Dec;45:355-367) (**Pediatr Neurol Briefs** 2012 Jan;26(1):3-4).

SEIZURE DISORDERS

SEIZURES AND EPILEPSY IN SOTOS SYNDROME

Clinicians from the Child Neurology Division, Sapienza University of Rome, and 7 other pediatric neurology centers in Italy report a series of 19 Sotos syndrome (SS) patients with febrile seizures (FS) and/or epilepsy during childhood and a long-term follow-up. Fifteen patients were male and 4 female. Mean age at first evaluation was 5 years 2 months (range 4 months–15 years). Minimum follow-up period was 5 years (range 5-19 years). FS were recorded in 11(58%) patients and afebrile seizures (AFS) in 15 (79%). FS were only simple in 6/11 (55%), only complex in 1 (9%), and both simple and complex in 4 (36%). AFS were temporal lobe type in 40%, and were responsive to AED therapy. All patients had the typical facial features of SS (high, broad forehead, down-slanting palpebral fissures, and pointed chin), tall stature and macrocephaly, and learning disability. Neurologic disorders associated with SS are hypotonia, incoordination, delayed language and motor development, behavioral abnormalities, and seizures. (Nicita F, Ruggieri M, Polizzi A, et al. Seizures and epilepsy in Sotos