development of WS, were not precursors of hypsarrhythmia. Given the importance of early treatment of infantile spasms with ACTH for optimal benefit, the definition of a reliable pre-hypsarrhythmia pattern in the EEG of infants at risk of developing West syndrome should lead to more effective therapy and improved prognosis.

STXBP1 mutations account for one-third of cases of early infantile encephalopathy with suppression-burst pattern (Ohtahara syndrome) (Saitsu H et al. **Epilepsia** Dec 2010;51(12):2397-2405). Also, STXBP1 mutation should be considered as a possible cause of symptomatic West syndrome without known cause. (Otsuka M et al. **Epilepsia** Dec 2010;51(12):2449-2452).

NEUROMUSCULAR DISORDERS

BENIGN COX DEFICIENCY MYOPATHY

Clinical and pathological features of benign infantile mitochondrial cytochrome c oxidase (COX) deficiency were studied in 8 patients with the disease phenotype seen at the National Center of Neurology and Psychiatry, Tokyo, Japan. Clinically, patients had severe generalized muscle hypotonia and weakness from birth to 3 months of age. Two patients had facial muscle involvement and high-arched palate. Prior to muscle biopsy and mitochondrial DNA analyses, including northern blot and respiratory chain enzyme activity assay, initial diagnoses were mitochondrial myopathy in 5 patients, congenital myopathy in 2, and congenital muscular dystrophy in 1. All patients except 1 recovered completely during a 3-year follow-up period. All learned to walk by 1-2 years of age and could run at follow-up. Blood lactate levels were normal in 2, slightly elevated in 4, and >twice normal level in 2. Serum CK was elevated in all infants (182-644 U/L) and was normal at follow-up. Brain MRI was normal at age 7 months and 13 months but showed abnormally high T2-weighted signals in bilateral caudate nuclei and putamina at ages 23 and 33 months in 2 siblings; MRI was normal in 2 other patients examined. Muscle biopsy showed variation in fiber size, and numerous ragged-red fibers that were COX negative. Northern blot analysis revealed decreased levels of mitochondrial transfer RNA-glutamate molecules. All patients had a novel homoplastic m.14674T>C or m.14674T>G mutation as the site of disease causation, resulting in defects of COX and multiple respiratory chain enzymes. In addition to muscle, the basal ganglia may also be involved. Normal respiratory chain enzyme activities found in naïve myoblasts was evidence of potential for spontaneous recovery. (Mimaki M, Hatakeyama H, Komaki H, et al. Reversible infantile respiratory chain deficiency: a clinical and molecular study. **Ann Neurol** Dec 2010;68(6):845-854). (Respond: Dr Y Goto, National Institute of Neuroscience, Kodaira, Tokyo 187-8502, Japan. E-mail: goto@ncnp.go.jp).

COMMENT. The authors conclude that the pathogenicity of the mutation at np 14674 in tRNA, resulting in deficiency of multiple respiratory enzymes, is confirmed. Compensatory mechanisms result in variation of clinical features from asymptomatic to both muscle and CNS involvement. They propose a change in name to "infantile reversible respiratory chain deficiency" since multiple respiratory chain enzyme deficiency may result in more than myopathy alone.