## LEVODOPA DOSE-LIMITING DYSKINESIAS IN TREATMENT OF SEGAWA DISEASE

Researchers at the University of Cordoba, and other centers in Spain, Germany and New York report an atypical phenotype of persistent, low-dose levodopa treatment-limiting dyskinesias in 2 members of a family (female child aged 13 years and mother) with prominent brachial dystonia and a novel GCH1 mutation. The girl proband developed severe writer's cramp at age 10 years and persistent treatment limiting dyskinesias, despite continuous levodopa therapy from age 7 years. Even low doses of levodopa (1.1-1.4 mg/kg/day) were associated with postural tremor and worsening of dystonia and writer's cramp. An improvement in dystonia followed an increase in the dose of levodopa to 2 mg/kg, but further increases resulted in dyskinesias, characterized by choreic movements and persistence of writer's cramp. The addition of trihexyphenidyl 6 mg/day and continued levodopa at 0.8 mg/kg/day controlled the dyskinesias. Dyskinesias as limiting side effects of levodopa should not preclude a diagnosis of dopa-responsive dystonia and Segawa disease. (Lopez-Laso E, Beyer K, Opladen T, Artuch R, Saunders-Pullman R. Dyskinesia as a limiting factor in the treatment of Segawa disease. **Pediatr Neurol** 2012 June;46:404-406). (Respond: Dr Lopez-Laso, Pediatric Neurology Unit, Department of Pediatrics, University Hospital Reina Sofia, Maimonides Institute of Biomedical Research of Cordoba, University of Cordoba, Av Menendez Pidal s/n, 14004 Cordoba, Spain. E-mail: elolaso@gmail.com).

COMMENT. Segawa disease is distinguished from *idiopathic torsion dystonia* (ITD) or Oppenheim's dystonia, autosomal dominant primarily affecting Ashkenazi Jewish families and presenting at about 10 years of age. ITD begins in the foot or arm and is progressive. *Dopa responsive dystonia* (DRD) or Segawa variant is autosomal dominant due to a mutation in the GCH-1 gene and deficiency of the enzyme guanosine triphosphate cyclohydrolase I. Diagnosis is by CSF analysis for reduced biogenic amines and pterins. DRD presents in early childhood, usually with foot dystonia, worse later in the day and better after sleep (diurnal variation in symptoms). DRD is usually responsive to small doses of levodopa, and levodopa-induced dyskinesias are typically absent. Some patients present with parkinsonism features. *Focal dystonias* (eg. blepharospasm, torticollis, writer's and musician's cramp), the most common forms of dystonia, typically affect women 40–60 years of age, but may also occur in children with dopa responsive dystonia. The pathophysiology of DRD is reviewed by Segawa M et al. **Ann Neurol** 2003;54 Suppl 6:S32-455).

# LEUKOENCEPHALOPATHIES

## MRI AND INFANTILE-ONSET LEUKOENCEPHALOPATHY

Researchers at VU University Medical Center, Amsterdam and other international centers reviewed the MRIs of >3000 patients with an unclassified leukoencephalopathy, and 7 patients (3 male) shared similar MRI abnormalities and clinical and laboratory