Infantile onset spinocerebellar ataxia. Identification of a novel Twinkle mutation is reported in a family with infantile onset spinocerebellar ataxia in two individuals manifesting ataxia, peripheral sensory neuropathy, athetosis, seizures, deafness, and ophthalmoplegia (Dundar H, et al. **Pediatr Neurol** 2012 Mar;46(3):172-7).

PATHOPHYSIOLOGY OF PRIMARY AND SECONDARY DYSTONIAS

Investigators at the Institute of Neurology, University College London, UK, and centers in Slovenia and Spain compared electrophysiological features of primary and secondary dystonia, using transcranial magnetic stimulation of motor cortex and eye blink conditioning. Eleven patients with hemidystonia secondary to basal ganglia or thalamic lesions were tested over both hemispheres, corresponding to the affected and non-affected sides, and compared with 10 patients with primary segmental dystonia with arm involvement and 10 healthy controls. All subjects were tested as adults. The average age at onset of secondary dystonia was 13.6 years (range 1–55; 5 were 1-2 years). The causes of secondary dystonia were perinatal HII in 5, ischemic stroke in 5, and encephalitis in 1.

No differences in motor thresholds were detected between patients with secondary and primary dystonia or controls. In secondary dystonia, short interval intracortical inhibition was reduced on the affected side, but normal on the non-affected side; cortical plasticity and eye blink classical conditioning were normal. In contrast, patients with primary dystonia showed increased cortical plasticity and reduced eye blink classical conditioning. Dystonia is a motor symptom that reflects different pathophysiological mechanisms. (Kojovic M, Parees I, Kassavetis P, et al. Secondary and primary dystonia: pathophysiological differences. **Brain** 2013 Jul;136(Pt 7):2038-49). (Response: Dr Maja Kojovic. E-mail: maja.kojovic.09@ucl.ac.uk).

COMMENT. **Adams and Victor's Principles of Neurology** (9th ed. New York: McGraw Hill Medical; 2009) lists 4 main groups of diseases characterized by dystonia: 1) *Hereditary and Degenerative* dystonias (dystonia musculorum deformans, Huntington chorea, Juvenile dystonia – Parkinson syndrome [L-dopa responsive]), 2) *Drug-induced* (phenothiazine, haloperidol), 3) *Symptomatic (secondary)* (Wilson disease, double athetosis cerebral palsy due to cerebral hypoxia, kernicterus), and 4) *Idiopathic focal dystonias* (spasmodic torticollis, blepharospasm, hemifacial spasm, oromandibular, spasmodic dysphonia, writer's cramp).

BIOTIN-RESPONSIVE OPHTHALMOPLEGIA / DYSTONIA

A 10-year-old girl with a 4-month history of abnormal gait and dysarthria had bilateral external ophthalmoplegia, dystonia, and altered mental status. MRI showed a characteristic "bat-wing" appearance and increased signal involving the medial nucleus of the thalamus, basal ganglia and cerebellum, suggesting biotin-responsive basal ganglia disease. Immediate improvement followed biotin and thiamine therapy. Repeat MRI showed resolution of vasogenic edema but residual atrophy and gliosis in the basal ganglia. The disease is autosomal recessive with SLC19A3 gene mutation, related to

thiamine transporter-2 deficiency. (Tabarki B, Al-Sheikh F, Al-Shahwan S, Zuccoli G. Bilateral external ophthalmoplegia in biotin-responsive basal ganglia disease. **J Pediatr** 2013 Jun;162(6):1291-2). (Response: Dr Saad Al-Shahwan, Prince Sultan Military Medical City, Riyadh, Saudi Arabia).

COMMENT. The authors propose that biotin-responsive basal ganglia disease be considered in a case of unexplained acute dystonia, external ophthalmoplegia, confusion and encephalopathy.

HEADACHE DISORDERS

DIAGNOSTIC CRITERIA FOR VESTIBULAR MIGRAINE

Investigators at the University of Pittsburgh, PA, review the latest clinical diagnostic criteria, pathophysiology, and treatment of vestibular migraine. Diagnosis requires all four of the following criteria:

- At least 5 episodes with vestibular symptoms lasting between 5min and 72h;
- Migraine with or without aura, present or previous history;
- One or more migraine features with at least 50% vestibular episodes;
- Not explained by another vestibular disorder.

Physical examination is generally normal between episodes. During episodes, nystagmus suggests a central or peripheral vestibular abnormality. Non-paroxysmal positional nystagmus is especially common. Vestibular migraine has a strong female preponderance, up to 5 to 1. Triggers are the same as those for migraine headache, including menstruation, sleep disorder, stress, physical exertion, dehydration, and food and drinks. Related disorders include Meniere's disease, benign paroxysmal positional vertigo, and anxiety. Treatment includes removal of triggers and pharmacotherapy, similar to that employed for migraine headache. The pathophysiology of vestibular migraine is incompletely understood. (Furman JM, Marcus DA, Balaban CD. Vestibular migraine: clinical aspects and pathophysiology. **Lancet Neurol** 2013 Jul;12(7):706-15). (Response: Prof. Joseph M Furman, University of Pittsburgh School of Medicine. E-mail: furmanjm@upmc.edu).

COMMENT. The association of recurrent vertigo with migraine in children was described as benign paroxysmal vertigo in 1964 (Basser L. **Brain** 1964 Mar;87:141-52). In adults, report of this association was delayed until 1984 (Kayan A, Hood JD. **Brain** 1984 Dec;107 (Pt 4):1123-42). Diagnostic criteria for vestibular migraine were published by the International Headache and Barany Societies in 2012 (Lempert T, Olesen J, Furman J, et al. **J Vestib Res** 2012;22(4):167-72).