panic attacks, and REM sleep behavior disorder. Epilepsies confused with sleep disorders include Rolandic epilepsy, Panayiotopoulos syndrome, occipital epilepsy of Gastaut, and nocturnal frontal lobe epilepsy (NFLE) in which seizures occur mainly during sleep. Guidelines are suggested for differentiating non-epileptic parasomnias from nocturnal seizures, and NFLE from arousal disorders. Non-convulsive status is sometimes misinterpreted as a sleep disorder. (Stores G. Sleep disturbance in childhood epilepsy: clinical implications, assessment and treatment. **Arch Dis Child** 2013 Jul;98(7):548-51). (Response: Professor Gregory Stores, Department of Psychiatry, University of Oxford, UK. E-mail: gregory.stores@psych.ox.ac.uk).

COMMENT. The author concludes that screening for sleep disturbance should be routine in children with epilepsy, and the accurate diagnosis determined, according to the **International Classification of Sleep Disorders: Diagnostic and Coding Manual** (2nd ed. Westchester, IL: American Academy of Sleep Medicine; 2005).

HEREDO-DEGENERATIVE / METABOLIC DISORDERS

CHARACTERISTICS OF INDIVIDUALS AT RISK FOR SPINOCEREBELLAR ATAXIA

Investigators at University Hospital of Bonn and 13 other centers in Germany, France, and Italy conducted a prospective, longitudinal observational study (2008-2011) of offspring or siblings of patients with spinocerebellar ataxias (SCA)-1, 2, 3, and 6. Study individuals had no ataxia and were aged 18-50 years (35-70 years, if directly related to individuals with SCA6). Relations between outcome variables and time from onset (present age to estimated age at ataxia onset) were analyzed using clinical scales, questionnaires, and coordination tests. In 264 participants, estimated time to ataxia from onset was -9 years in 50 carriers of the SCA1 mutation, -12 years in 312 SCA2 mutation carriers, -8 years in 26 SCA3 mutation carriers, and -18 years in 16 SCA6 mutation carriers. Compared with non-carriers of each mutation, SCA1 and SCA2 mutation carriers had higher median scores for ataxia. SCA2 carriers had lower functional index scores than did non-carriers and worse composite cerebellar functional scores than did their non-carrier counterparts. In 83 individuals (30%) who underwent MRI, brainstem and cerebellum showed grey-matter loss in SCA1 and SCA2 mutation carriers. (Jacobi H, Reetz K, Montcel ST, et al. Biological and clinical characteristics of individuals at risk for spinocerebellar ataxia types 1, 2, 3, and 6 in the longitudinal RISCA study: analysis of baseline data. **Lancet Neurol** 2013 Jul;12(7):650-8). (Response: Prof T Klockgether. E-mail: klockgether@uni-bonn.de).

COMMENT. The authors summarize the baseline findings of the RISCA study designed to define the preclinical stages of SCAs in individuals at risk and to identify functional and brain structural abnormalities before the onset of ataxia. Carriers of SCA1 and SCA2 mutations have mild coordination functional deficits and brain structural abnormalities before the onset of clinically manifest ataxia. Recognition of the preclinical manifestations of neurodegenerative diseases may permit early therapeutic intervention.

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