Approach to the diagnosis of neurotransmitter diseases exemplified by the differential diagnosis of childhood-onset dystonia. Ann Neurol 2003;54(suppl 6):S18-S24). (Respond: Dr Assmann, University Children's Hospital, Moorenstrasse 5, 40225 Dusseldorf, Germany).

COMMENT. The term dystonia is used for a symptom or abnormal movement disorder. Dystonia is characterized by intermittent or continuous muscle spasms that are generalized (dystonia musculorum deformans) or segmental (localized to the neck (spasmodic torticollis), upper limb, or lower limb), and primary genetic or acquired (post-perinatal asphyxia, trauma, toxins, or vascular). In primary hereditary dystonias, the mode of transmission is variable. One form with high incidence in Ashkenhazi Jews is probably autosomal dominant. The first symptom in hereditary dystonia is an involuntary posturing of one portion of the body, most commonly a plantar, flexion-inversion movement of the foot, commonly misdiagnosed as a hysterical gait. In treatment, trihexyphenidyl (Artane) is used in Ashkenazi Jewish patients, and Levodopa, alone or with a decarboxylase inhibitor (Sinemet), in patients with late onset dystonia. Cryothalamectomy is a surgical approach rarely entertained in children with medically-intractable hemidystonia.

CSF analysis and magnetic resonance spectroscopy in the diagnosis of neurotransmitter diseases are reviewed by Hyland K (Ann Neurol 2003;54(suppl 6):S13-S17), and Novotny EJ Jr et al (Ann Neurol 2003;54(suppl 6):S25-S31). The CSF compounds measured are homovanillic acid (end product for dopamine metabolism), 5-hydroxyindoleacetic acid (for serotonin), and 3-methoxy-4-hydroxyphenylglycol (for norepinephrine). MRS methods are under investigation for the measurement of neurotransmitters in the brain. Abnormalities of motor cortex excitability preceding voluntary movement in patients with dystonia have been studied by Gilio F et al (Brain 2003;126:1745-1754). Dystonic movements are commonly triggered or made worse by voluntary action.

PAROXYSMAL DYSKINESIAS

Clinical characteristics of 26 children diagnosed with paroxysmal dyskinesias between 1980 and 2000 were evaluated retrospectively at the National Neurological Institute "C Besta" of Milan, Italy. Patients were categorized according to precipitating factors: 14 had paroxysmal kinesigenic dyskinesia (PKD), 6 had paroxysmal non-kinesigenic dyskinesia (PNKD), and 6 had paroxysmal exercise-induced dyskinesia (PED). None had paroxysmal hypnogenic dyskinesia (PHD), a form of nocturnal frontal lobe epilepsy. Patients with PKD had a mean age at onset of 7.1 years (range1.5-14 years); 13 were idiopathic, with a positive family history in 9 and autosomal-dominant inheritance; and one was associated with Chiari type 1 malformation. Of the 6 with PNKD, 1 had multiple sclerosis, 2 had cerebral palsy, 1 had a left basal ganglia stroke. 1 an acute inflammatory encephalopathy, and only 1 was idiopathic. Six with PED were all idiopathic, and attacks of dystonia or choreoathetosis were triggered by prolonged exercise, usually running or walking. Antiepileptic drugs, especially carbamazepine, were most effective in treatment of the PKD type, with benefit obtained in 70%. The occasional co-occurrence of epilepsy and PKD may be explained by a common ion channel dysfunction, and a gene locus mapped to chromosome 16. (Zorzi G, Conti C, Erba A et al. Paroxysmal dyskinesias in childhood. Pediatr Neurol March 2003;28:168-172). (Respond: Giovanna Zorzi MD, Department of Child Neurology, Instituto Nazionale Neurologico "C Besta", Milano, Italy).

COMMENT. Paroxysmal dyskinesias refer to brief attacks of dystonia or choreoathetosis, alone or combined, with return to normal between episodes. Cases are classified according to precipitating factors. Paroxysmal kinesthetic dyskinesia (PKD) is the most frequent form of paroxysmal dyskinesia, it is usually familial and idiopathic, often responds to treatment with carbamazepine, and may resolve spontaneously by age 30. PNKD is relatively rare, it is often symptomatic of illnesses including cerebral palsy, infection, or vascular lesion, and response to treatment is poor. Exercise-induced (PED) cases are rare, usually in response to running, and dystonia is generalized or focal, affecting the foot. Antiepileptic drugs, including carbamazepine, clonazepam, and acetazolamide, L-Dopa, and trihexyphenidyl are reported of benefit in some cases.

LANGUAGE DISORDERS

LANGUAGE REGRESSION, AUTISM, AND EPILEPSY

The records of 196 consecutive children (143 males and 53 females) with language regression evaluated between 1988 and 1994 by a child neurologist were analyzed at the Department of Neurology, Albert Einstein College of Medicine, Bronx, NY. Patients with a neurodegenerative or metabolic disorder, acquired brain injury, or Rett syndrome were excluded. Mean age at regression, defined as loss of previously acquired language skills, was 21.2 months (SD 10.5), the mean age at first visit was 50.6 months (SD 41.1), and the mean interval between onset and referral was delayed for 34.8 months (SD 38.3). Trigger factors that preceded the onset of language regression were reported in 74 (38%); these were definite in 34 (17%) and possibly related in 40 (20%). They included illness (23%), emotional upset (16%), seizures (3%), and trauma (1%). A history of seizures was present in 15%, but seizures were temporally related to language regression in only 3%. Repeated ear infections were the most frequently reported abnormality in the past history, occurring in 32%. At the time of neurologic examination, autistic spectrum disorder was diagnosed in 93%, and cognitive impairment occurred in 73%, with severe mental retardation in 3%. Hearing was normal in 94%, and mildly impaired in 6%. Stereotypies were noted in 85%, hypotonia in 58%, toe walking in 15%, and oromotor deficits in 11%. Sensorimotor development was normal in 29%. CT/MRI obtained in 75 children was abnormal in only 3 (4%). Chromosomes and metabolic tests were normal in all 47 tested. EEGs showed paroxysmal abnormalities in 4 of 12 (33%) with prolonged video monitoring. At follow-up (mean age 64 months) 39% were nonverbal and an additional 45% showed decreased verbal output. Some recovery occurred in 61% but only one child recovered fully. Improvement was most likely in the 49% who were developmentally normal before language regression was noted. (Wilson S, Djukic A, Shinnar S et al. Clinical characteristics of language regression in children. Dev Med Child Neurol August 2003;45:508-514). (Respond: Isabelle Rapin MD, Albert Einstein College of Medicine, K807, 1300 Morris Park Ave, Bronx, NY 10461).