Late presentation of biotinidase deficiency is described in a previously healthy 5-year-old girl who developed acute visual loss and optic atrophy, and an ataxic gait. Classical signs of biotinidase deficiency were absent. (Rahman S, Standing S, Dalton RN, Pike MG. Dev Med Child Neurol Dec 1997;39:830-831).

Biotin deficiency and chronic anticonvulsant therapy. Nine adults treated with various anticonvulsants, including phenytoin and carbamazepine, compared to 17 controls showed a twofold increase in the 24-hour urinary excretion of bisnorbiotin, biotin sulfoxide, and 3-hydroxyisovaleric acid, metabolites of biotin, whereas urinary and serum biotin concentrations were unchanged. Long-term treatment with anticonvulsants may be associated with an increased biotin catabolism. (Mock DM, Dyken ME. Neurology Nov 1997;49:1444-1447).

DEGENERATIVE DISEASES

MITOCHONDRIAL DNA MUTATION IN RETT SYNDROME

Analysis of mitochondrial DNA from 15 children with Rett syndrome (RS) and 14 of their mothers is reported from the Department of Pediatrics, Beijing Medical University, China. Polymerase chain reaction amplification and single strand conformation polymorphism analysis showed mutations in region 2650-3000 encoding 16S rRNA of mtDNA in 13 patients with RS and 11 mothers. DNA sequence analysis and mismatch PCR results confirmed a point mutation (C --> T) at position 2835 in 7 patients with RS and in 6 of their mothers, that was absent in controls. (Tang J, Qi Y, Bao X-H, Wu X-R. Mutational analysis of mitochondrial DNA of children with Rett syndrome. Pediatr Neurol Nov 1997;17:327-330). (Respond: Dr Xi-Ru Wu, Department of Pediatrics, The First Teaching Hospital, Beijing Medical University, Beijing 100034, PR China).

COMMENT. Most cases of Rett syndrome are sporadic, but a few familial examples are reported. A maternal inheritance pattern suggests that mitochondrial DNA may be involved. The mutations observed in the mtDNA of patients with Rett syndrome and their mothers lends support to the hypothesis of a genetic basis for the disorder in some cases.

Japanese monozygotic female twins with Rett syndrome are reported from Fukuoka University, Japan. (Ogawa A, Mitsudome A, Yasumoto S, Matsumoto T. Brain Dev Dec 1997;19:568-570). The two 28-year-old patients had discordant characteristics regarding seizures, scoliosis, and stereotypic hand movements in adolescence. The authors cite 7 pairs of monozygotic twins with RS reported in the literature, and 11 pairs of dizygotic twins, only one twin affected, always the female.

CSF SUBSTANCE P LEVELS IN RETT SYNDROME

The cerebrospinal fluid (CSF) levels of neuropeptide substance P were measured in 20 patients with Rett syndrome and controls at Kurume University, Japan, and other centers. CSF substance P levels are constant in control children between 2 and 12 years, and show a gradual decrease through adolescence, reaching a plateau at 20 years. Significant reductions in substance P in patients with RS compared to controls were present at the early phases of the disease, at age 2 to 3 years, and were not age dependent. Childhood RS levels were 50% of controls in the same age group, and adults with RS had 37% of control adult

values. The mean CSF levels of substance P in patients with mental retardation, epilepsy, and Guillain-Barre disease were not different from controls without neurologic disease. (Matsuishi T, Nagamitsu S, Yamashita Y et al. Decreased cerebrospinal fluid levels of substance P in patients with Rett syndrome. Ann Neurol Dec 1997;42:978-981). (Respond: Dr Matsuishi, Department of Pediatrics and Child Health, Kurume UIniversity School of Medicine, 67 Asahi-machi, Kurume City, Japan 830).

COMMENT. CSF concentrations of substance P have been shown to reflect brain and spinal cord concentrations. The authors suggest that decreased CSF levels of substance P in patients with RS, if confirmed in other studies, may be useful as a biological marker for the disease.

SPEECH AND LANGUAGE DISORDERS

ACQUIRED CHILDHOOD DYSARTHRIA CLASSIFICATION

Published reports of acquired childhood dysarthria since 1980 were reviewed at University Hospital, Rotterdam; and the Department of Medical Psychology, Ziekenhuis Walcheren, Vlissingen, The Netherlands, and cases were classified on the basis of neuroradiological location of lesion and associated motor disorders. The majority of cases (20) were examples of mutism and subsequent dysarthria (MSD) following resection of cerebellar tumor. Slow articulation, monotony, and hoarse soft voice were the most frequent manifestation of dysarthria, and all patients had severe limb and trunk ataxia. Recovery or improvements in speech and motor disability were dissociated, dysarthria resolving first and almost completely while ataxia often persisted. Cerebellar MSD in children is different from the adult form of ataxic dysarthria, which is characterized by excess and equal stress resulting in scanning speech. Scanning speech was rarely observed in children. In 5 children with basal ganglia lesions and extrapyramidal movement disorders, dysarthria was characterized by hypophonia, stuttering, and difficulty in controlling rate of speech, similar to the hypokinetic dysarthria in adults. Acquired dysarthria in childhood requires a separate classification from that of adults. (van Mourik M, Catsman-Berrevoets CE, Paquier PF, Yousef-Bak E, van Dongen HR. Acquired childhood dysarthria: Review of its clinical presentation. Pediatr Neurol Nov 1997;17:299-307). (Respond: Dr van Mourik, Department of Medical Psychology/Ziekenhuis Walcheren, Postbus 3200, 4380 DD Vlissingen, The Netherlands).

COMMENT. Acquired childhood dysarthria is classified as 1) ataxic type, usually following mutism as a complication of cerebellar tumor resection; and 2) hypokinetic type associated with basal ganglia lesions and extrapyramidal movement disorders.

The syndrome of cerebellar mutism and subsequent dysarthria is covered in Progress in Pediatric Neurology III, PNB Publishers, 1997;pp306-307. A report from University Hospital, Rotterdam cites 36 cases in the literature, including 5 of 15 children operated for cerebellar tumor, mainly medulloblastoma, at that hospital. The mutism was correlated with adherence of the tumor to the floor of the fourth ventricle. The dysarthria that followed lasted for 1 to 5 weeks.

SEIZURE DISORDERS

PREVALENCE OF LENNOX-GASTAUT SYNDROME IN ATLANTA

The prevalence and epidemiology of Lennox-Gastaut syndrome (LGS)