COMMENT. Patients with Down syndrome often suffer from thyroid disorders and congenital hypothyroidism is much more common in DS than in normal individuals. It is likely that the congenital hypothyroidism in the above patient was related to the chromosome 21 abnormality.

The clinical manifestations of partial deletion of chromosome 21 have included mild mental retardation, short stature, obesity, hypotonia, prominent forehead, downslanting palpebral fissures, hyperopia, large/low set ears, high arched palate, prognathism, long/slender hands, short 5th finger, broad feet, large stiff joints, and congenital hypothyroidism.

MENTAL RETARDATION SYNDROMES

SMITH-LEMLI-OPITZ SYNDROME

Clinical features as specific indicators in the diagnosis of Smith-Lemli-Opitz syndrome (SLOS) and the reliability of ultraviolet spectrophotometry (UVS) as a biochemical screening test were examined by an Italian SLOS Collaborative Group of investigators. Of 20 patients with clinical suspicion of SLOS, referred to 11 Italian pediatric and clinical genetic centers in 1994, the diagnosis was confirmed biochemically by gas chromotography/mass spectrometry analysis (GC/MS) of serum sterols in 10, and serum sterols were normal in 10. Comparison of clinical signs in confirmed cases and biochemically negative patients did not reveal a specific group of manifestations of SLOS. UVS measurement of 7-dehydrocholesterol, which accumulates in the plasma in SLOS, correlated with GC/MS profiles. Serum bile acid concentrations were lower than normal in 4 of 5 patients with the syndrome. (Guzzetta V, Andria G et al. Clinical and biochemical screening for Smith-Lemli-Opitz syndrome. Acta Paediatr Aug 1996;85:937-942). (Respond: Dr G Andria, Department of Pediatrics, Federico II University, Via Pansini 5, 80131 Naples, Italy).

COMMENT. The "gestalt" impression formed by an experienced clinician examining the facial appearance of a child is perhaps the most practical and reliable method of diagnosis of Smith-Lemli-Opitz syndrome. Signs and symptoms of the syndrome are variable and non-specific and include mental retardation, failure to thrive, feeding difficulties, hypotonia, microcephaly, ptosis and epicanthal folds, anteverted nostrils, micrognathia, low set ears, syndactyly, simian creases, and hypospadias. Ultraviolet spectrophotometry determination of serum 7-DHC levels is 100% sensitive, relatively inexpensive, and specific for the biochemical diagnosis of SLOS.

PSYCHIATRIC DISORDERS IN MENTALLY RETARDED, EPILEPTIC CHILDREN AND ADOLESCENTS

The prevalence and types of psychiatric disorders in 98 school-age children with mental retardation (MR) and active epilepsy were investigated in the Departments of Child and Adolescent Psychiatry and Pediatrics, University of Goteborg, Sweden. At least 1 psychiatric diagnosis was uncovered in 53 (59%) patients, and symptoms could not be classified because of profound MR in 30 (33%). Autistic disorder was diagnosed in 24 (27%), and autistic-like disorder in 10 (11%). ADHD was present in 11, and Angelman syndrome in 4. In those with autism the most common seizures were complex partial, absence, myoclonic, and tonic-clonic. A history of infantile spasms occurred in 12

(13%). Many of the psychiatric disorders had not previously been diagnosed despite parental concern. (Steffenburg S et al. Psychiatric disorders in children and adolescents with mental retardation and active epilepsy. Arch Neurol Sept 1996;53:904-912). (Respond: Dr Suzanne Steffenburg, Department of Child and Adolescent Psychiatry, University of Goteborg, Annedals Clinic, S-413 45 Goteborg, Sweden).

COMMENT. Autism or autistic-like disorder are common in children with mental retardation and epilepsy and are frequently undiagnosed. Neurologists and psychiatrists might work in closer collaboration for optimal management of these patients.

SEIZURE DISORDERS

FAMILIAL TEMPORAL LOBE EPILEPSY IN TWINS

A new syndrome of familial temporal lobe epilepsy is described in 38 subjects from 13 unrelated families and was first identified in 5 concordant monozygotic twin pairs at the Australian National Health and Medical Research Council Twin Registry, University of Melbourne, Parkville, Australia. Seizure types were simple partial seizures with psychic or autonomic symptoms, infrequent complex partial seizures, and rare secondarily generalized seizures. EEGs showed focal temporal interictal epileptiform discharges in 22%. MRIs were normal. Autosomal dominant inheritance with age-dependent penetrance was likely. Some family members were affected with only mild and subtle seizure manifestations. (Berkovic SF et al. Familial temporal lobe epilepsy: A common disorder identified in twins. Ann Neurol Aug 1996;40:227-235). (Respond: Dr Samuel F Berkovic, Department of Neurology, Austin and Repatriation Medical Centre, Heidelberg (Melbourne), Victoria 3084, Australia).

COMMENT. Onset of familial temporal lobe epilepsy (TLE) is typically in adolescence or early adult life, whereas TLE with hippocampal sclerosis (HS) usually begins in childhood. Febrile seizures, often preceding the TLE of HS, were not increased in frequency in family members of familial TLE subjects. The mild and subtle nature of familial TLE may explain the previous infrequent reports of similar syndromes. Bray PF and Wiser WC have described the hereditary characteristics of familial temporo-central focal epilepsy, and the above authors suggest that some of their cases persisting into adulthood might represent examples of familial TLE. (Pediatrics 1965;36:207-211).

UNPROVOKED SEIZURES WITH FEBRILE SEIZURES

Unprovoked seizures occurred in 26 (6%) of 428 children followed for 2 years or more after a first febrile seizure at the Montefiore Medical Center, Bronx, NY. Risk factors for unprovoked seizures were neurodevelopmental abnormalities, complex febrile seizures, family history of epilepsy, recurrent febrile seizures, and a briefer duration of fever before the initial febrile seizure. Family history of febrile seizures, temperature and age at the initial febrile seizure were not associated risks for unprovoked seizures. (Berg AT, Shinnar S. Unprovoked seizures in children with febrile seizures: Short-term outcome. Neurology Aug 1996;47:562-568). (Reprints: Dr Anne T Berg, Social Science Research Institute, Northern Illinois University, DeKalb, IL 60115).