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