levels correlated directly with hyperactivity. (Rovet J, Alvarez M. Thyroid hormone and attention in school-age children with congenital hypothyroidism. J Child Psychol Psychiat July 1996;37:579-585). Reprints: Dr Joanne Rovet, Psychology Department, The Hospital for Sick Children, 555 University Ave, Toronto, Ontario, Canada M5G 1X8).

COMMENT. Higher levels of T4 and TSH, consistent with a resistance to thyroid hormone, are associated with poorer attention and less hyperactive behavior in children with congenital hypothyroidism. Children with CH should be closely monitored to maintain levels of T4 and TSH within the normal range and to avoid elevations of hormone that could impede attention. Thyroxine and thyrotropin have unique effects on specific aspects of attention and behavior.

The incidence of ADHD is 10 times higher in children with thyroid hormone resistance than in those with normal thyroid function. (Hauser P et al. N Engl J Med 1993;328:997). See Progress in Pediatric Neurology II, 1994, pp 177-8. Studies of thyroid function should be included more frequently in the clinical evaluation of children with ADHD.

THYROXINE-INDUCED ABSENCES IN JME

A patient with juvenile myoclonic epilepsy (JME), manifested as absences at age 10 and myoclonic jerking and generalized tonic clonic seizures at age 15, was treated successfully with primidone at the King Fahad National Guard Hospital, Riyadh, Saudi Arabia. Absence seizures were provoked by thyroxine 50 mcg daily prescribed at age 35 for a simple goitre associated with normal thyroid function tests. Loss of concentration lasting a few seconds was associated with frequent discharges of sharp, spike, multispike, and slow wave complexes on the EEG. The thyroid levels were slightly elevated compared to initial tests. Absences stopped and the EEG returned to normal after thyroxine was discontinued. (Obeid T et al. Thyroxine exacerbates absence seizures in juvenile myoclonic epilepsy. Neurology Aug 1996;47:605-606). (Respond: Dr Tahir Obeid, Division of Neurology, King Fahad National Guard Hospital, PO Box 22490, Riyadh 11426 Saudi Arabia).

COMMENT. When thyroid hormone is administered in patients with epilepsy, the dose should be monitored carefully and an EEG obtained to examine for subclinical absence seizures requiring additional antiepileptic medication.

HYPOTHYROIDISM AND CHROMOSOME 21 DELETION

A 12-year-old mentally retarded girl with a large deletion of the long arm of chromosome 21 and congenital hypothyroidism is reported from the Department of Clinical Genetics and Pediatrics, University Hospital, Uppsala, Sweden. Despite the large chromosome deletion, the degree of mental retardation was mild and severe manifestations of Down syndrome were absent. The proximal part of the long arm of chromosome 21 does not include the genes responsible for severe clinical effects seen in Down syndrome. Congenital hypothyroidism in this patient might indicate the importance of genes on chromosome 21 for thyroid function. (Ahlbom BE, Sidenvall R, Anneren G. Deletion of chromosome 21 in a girl with congenital hypothyroidism and mild mental retardation. Am J Med Genet Aug 1996;64:501-505). (Reprints: Dr G Anneren, Department of Clinical Genetics and Pediatrics, University Hospital, S-751 85 Uppsala, Sweden).

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