deficiency of holocarboxylase synthetase and 2. late infantile disease due to biotinidase deficiency. Neurological manifestations are prominent in the late-onset group and seizures may precede the cutaneous eruption and loss of hair.

Having encountered one such patient who presented at the age of 5 years with ataxia and seizures refractory to medications (In Nutrition, Diet, and your Child's Behavior. Charles C Thomas, Springfield, 1986.), I now make it a practice to prescribe biotin 10 mg daily as a therapeutic test when this diagnosis is suspected. Treatment reverses the organic aciduria so that a urine collection for analysis should precede administration of the vitamin. A dramatic response to a vitamin in a single daily dose is certainly an improvement over long-term anticonvulsant drug therapy with its attendant potential side-effects. Supplies of biotin from Roche Labs are available only for research at present (See Biotin. Ed. by Dakshinamurti K, Bhagavan HN. Ann. N.Y. Acad Science. New York, 1985; 447, 222-224, 297-313)

TUBEROUS SCLEROSIS AND INFANTILE SPASMS

Forehead plaques, smooth patches of slightly raised skin with a reddish or yellowish discoloration, can be the earliest skin manifestation of tuberous sclerosis (TS) according to the authors who describe 2 patients seen at Bath and Bristol, UK., presenting with infantile spasms at 3 and 5 months of age. (Fryer AE et al. Arch Dis Child 1987; 62:292-293)

COMMENT: Early diagnosis of tuberous sclerosis (TS) is important for genetic counselling and prognostic predictions. The prevalence of TS in patients with infantile spasms has been estimated at 25% or higher in some series. A Wood's light examination of the skin for hypopigmented maculae, a more frequent characteristic dermatologic manifestation of TS, is important in all infants with myoclonic spasms and hypsarrhythmia.

THYROTROPIN-RELEASING HORMONE (TRH): AN ALTERNATIVE THERAPY FOR INFANTILE SPASMS

Pediatric neurologists at the Central Hospital, Aichi Prefectural Colony, Kasugai, Aichi 480-03, Japan, compared the effects of TRH in 31 children and ACTH in 33 with severe epilepsy. Approximately half the cases had infantile spasms and the remainder had Lennox-Gastaut syndrome. In the TRH group, complete control of infantile spasms occurred in 7 of 13 (53.7%) and marked improvement of the EEG's was observed in 8 (61.5%). In the ACTH group, infantile spasms were controlled in 75%. TRH treated patients had no serious side-effects whereas 66.7% of the ACTH group had complications, including pneumonia, hypokalemia, cataracts, and brain shrinkage.

TRH-tartrate (TRH-t), 0.5 - 1.0 mg, was administered intravenously to determine immediate effects on seizures and EEG and then intra-muscularly once daily for 1 - 4 weeks. TRH was effective in controlling infantile spasms within 4 - 16 days of its initiation. Three of the 7 responders remained seizure-free for > 6 months. (Matsumoto A, Kumagai T, Takenchi T, Miyazaki S, Watanabe K. Epilepsia 1987;28:49-55)

COMMENT: ACTHis effective in the control of infantile spasms and hypsarrhythmia in 50% of cases. The response rate is higher in infants treated early and under one year of age than in those diagnosed later. Hypertension, cushingnoid obesity, congestive heart failure, infection, and cerebral atrophy are some of the more serious side-effects of ACTH therapy. The significant response of infantile spasms to TRH without serious toxicity offers a promising alternative therapy to ACTH. The anticonvulsant action of TRH appears to be central and unrelated to its endocrine action through the pituitary-thyroid axis.

NEOPLASMS AND RELATED CONDITIONS

NEUROFIBROMATOSIS

LINK (Let's Increase Neurofibromatosis Knowledge), the British Neurofibromatosis Association, organised a major European Symposium at Egham, Surrey, Feb 5-7, 1987, and clarified the distinguishing features of two syndromes with separate genetic markers: 1) von Recklinghausen's neurofibromatosis (VRNF), the so-called peripheral type, and 2) bilateral acoustic neurofibromatosis (BANF), the central variety.

VRNF with a prevalence of 1 in 3000 is inherited as an autosomal dominantcondition with 100% penetrance and a high mutation rate. Serious complications, occurring in about 20% include large plexiform neurofibromas, kyphoscoliosis, and optic nerve or chiasmal gliomas. Children should be examined twice a year to check for complications.

The gene responsible for VRNF, although not identified, was narroweddown to a few chromosomes by data that provided an 'exclusion map' at this conference. The genetic analysis of BANF patients has shown deletions on chromosome 22. a step closer to the identification of the defective gene responsible for acoustic neuromas. (Lancet 1987; i:663-664)

COMMENT: A similar conference on neurofibromatosis is scheduled for July 13 - 15, 1987 in the U.S. to be sponsored by the National Institute of Health, Bethesda, Md. and chaired by Dr. David A. Stumpf of Northwestern University Medical School. It is perhaps unfortunate that the European and US sponsors could not have pooled their resources to make this an International Symposium.

DEGENERATIVE DISORDERS

RETT'S SYNDROME

The sleep and respiratory patterns associated with this disorder have been studied in 11 females aged 2 through 15 years at the Methodist Hospital, Houston, Tx. Polygraphic recordings showed a pattern of disorganised breathing and compensatory hyperpnea during wakefulness with regular, continuous breathing during sleep. The findings suggest an altered or impaired voluntary/behavioural respiratory control system in patients with Rett's Syndrome. (Glaze DG, Frost JD Jr, Zoghbi HY, Percy AK. Rett's Syndrome: characterisation of respiratory patterns and sleep. Ann Neurol 1987;21:377-382)