

COMMENT: ACTH is 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, cushingoid 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 dominant condition 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 narrowed down 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)