

injury and reduced NAA. (Kaufmann WE, Wang PY, Koth CW, Denckla MB, Barker PB. Evaluation of brain metabolism in neurofibromatosis type 1 by magnetic resonance spectroscopy. Ann Neurol September 1999;46:528. CNS 28th Annual Meeting Abstract, Oct 13-16, 1999).

COMMENT. This work is an extension of previous reports from Johns Hopkins University concerning the relationship between UBOs and lower IQs in children with neurofibromatosis-1 (Denckla MB et al. Am J Med Genet 1996;67:98-102). (see Progress in Pediatric Neurology III, PNB Publ, 1997;pp291-294, for a review of these and other reports, some showing conflicting results).

Ped Neur Briefs Nov 1997;11:84, reviews an article on learning disability subtypes in children with neurofibromatosis; academic underachievers fall into 3 groups: 40% have normal IQ test results, 50% have general learning disabilities, and 14% have visuospatial and motor coordination problems, without language deficits.

HYPOMELANOSIS OF ITO: A GENETIC MOSAICISM

Evidence purporting that the so-called hypomelanosis of Ito (HI) syndrome does not exist as a distinct multisystem birth defect is presented by geneticists and dermatologists at Bad Salzschlirf, and Philipp University of Marburg, Germany. HI is a nonspecific pigmentary disorder representing a cutaneous marker of many different states of genetic mosaicism. The clinical findings are highly variable, not always involving brain, eyes or bones, occurrence is sporadic, and cytogenetic abnormalities involve many different chromosomes, especially the X-chromosome. The terms HI, incontinentia pigmenti achromians, pigmentary dysplasia, and pigmentary mosaicism are synonyms of the same cutaneous signs. "Pigmentary mosaicism of the Ito type" should be substituted for the term "HI syndrome." (Kuster W, König A. Hypomelanosis of Ito: No entity, but a cutaneous sign of mosaicism. Am J Med Genet Sept 1999;85:346-350). (Respond: Wolfgang Kuster MD, Clinical Genetics, TOMESA Clinic for Allergy, Riedstraße 18, D-36361, Bad Salzschlirf, Germany).

COMMENT. HI is a relatively common disorder in pediatric neurology clinics, involving 1 in every 1000 patients attending a service in Spain (Pascual-Castroviejo I et al. Hypomelanosis of Ito; neurological complications in 34 cases. Can J Neurol Sci 1988;15:124-129). CNS anomalies include mental and motor retardation, microcephaly, hypotonia, hyperkinesia, ataxia, seizures, and deafness. Eye defects include microphthalmia, ptosis, nystagmus, cataracts, and retinal degeneration. Bone anomalies include dental enamel defects, short stature, limb asymmetry, scoliosis, syndactyly, and polydactyly. These multisystem defects are explained, not as a single syndrome, but by different genetic defects and a sign of mosaicism, as evidenced by a variety of reported underlying chromosomal abnormalities.

MOVEMENT DISORDERS

TOURETTE SYNDROME AND DYSTONIA: GENETICALLY RELATED

A three-generation family in which 5 members were diagnosed with focal dystonia and 3 with tics, Tourette syndrome, and hyperactivity is reported from the Radcliffe Infirmary, Oxford, and Queen Elizabeth Hospital, Birmingham, England. One with dystonic head tremor subsequently died of a motor neuron disease. The findings of dystonia and Tourette syndrome in 8 members of a single