NEUROCUTANEOUS SYNDROMES

FREQUENCY OF COGNITIVE DEFICITS IN NEUROFIBROMATOSIS

The frequency and severity of specific cognitive deficits in 81 children with neurofibromatosis type 1 (NF1), ages 8 to 16 years, compared to 49 unaffected sibling controls, were assessed in a study at the University of Sydney, New South Wales, Australia. Specific learning disabilities, defined by IQ-achievement discrepancies, occurred in 20% (37% in males and 5% females), but 51% showed impairments in reading, spelling, and mathematics, and 81% had moderate to severe impairments in cognitive functioning. Mental retardation was present in 6 – 7%. Attention deficit hyperactivity disorder was diagnosed in 38%, while 63% had sustained attention difficulties. Compared with their siblings, ADHD was 3 times more common in NF1 children, with similar frequencies in males and females. An NF1 neuropsychological profile shows deficits in visuo-spatial and –perceptual skills, executive functioning, attention, and expressive and receptive language. Verbal and visual memory was preserved. (Hyman SL, Shores A, North KN. The nature and frequency of cognitive deficits in children with neurofibromatosis type 1. **Neurology** October (1 of 2) 2005;65:1037-1044). (Reprints: Dr KN North, Children's Hospital at Westmead, Clinical Sciences Building, Locked Bag 4001, Westmead, NSW 2145, Australia).

COMMENT. A neuropsychological profile for neurofibromatosis type 1 is characterized by weaknesses in visuospatial and visuoperceptual skills and strengths in verbal and visual memory. Specific learning disabilities are particularly prevalent in males, and girls are relatively spared. Comorbidity with ADHD is prevalent, both in males and females with NF1.

COGNITIVE AND BEHAVIORAL DEFICITS IN NEUROCUTANEOUS SYNDROMES

Cognitive and behavioral features of Sturge-Weber syndrome, tuberous sclerosis, and neurofibromatosis are summarized by a literature review (113 references) at the New York University, New York. *Sturge-Weber syndrome* is associated with mental retardation in 50 to 60% of cases, correlated with the extent of unilateral cerebral calcification and atrophy. The occurrence of seizures in up to 90% of cases is associated with leptomeningeal angiomatosis, and predicts a poorer prognosis. Hemispherectomy provides an 80% rate of seizure freedom; mean age of surgery was 2 years for patients left with a mild disability compared to 3 years for those with moderate or severe disability. A progressive neurological and developmental deterioration may be explained by venous occlusions and hypoxia, and is sometimes correlated with a worsening of seizures and EEG abnormalities. Psychiatric symptoms in Sturge-Weber syndrome include irritability, social problems, ADHD, oppositional defiance disorder, self-abuse, aggressive behavior, and depression, often correlated with occurrence of seizures.

Tuberous sclerosis (TS) presents with seizures in 80 – 90% of cases, often developing in the first year of life, and manifesting as infantile spasms in one-third. An IQ below 70 was