PEDIATRIC NEUROLOGY BRIEFS

A MONTHLY JOURNAL REVIEW

J. GORDON MILLICHAP, M.D., F.R.C.P., EDITOR

Vol. 3, No. 12 December 1989

METABOLIC AND DEGENERATIVE DISORDERS

ADRENOLEUKODYSTROPHY

The biochemical and clinical efficacy of dietary erucic acid (C22:1) therapy for X-linked adrenoleukodystrophy (ALD) was investigated at the Departments of Pediatrics, Human Genetics, Neurology, Medical College of Virginia, Virginia Commonwealth University, Richmond, VA, and the Department of Neurology, University of California, Davis, Sacramento, CA. Twelve patients, aged six to 12 years, were treated with a diet enriched with erucic acid and oleic acid for 2-19 months. The mean plasma C26:0 concentration decreased to normal by four weeks, and the C26:0 composition of plasma sphingomyelin and phosphatidylcholine became normal by four months. Two mildly affected patients remained clinically stable whereas six with moderate to advanced disease deteriorated. The diet may prevent further demyelinization in some mildly affected boys with X-linked adrenoleukodystrophy. (Rizzo WB et al. Dietary erucic acid therapy for X-linked adrenoleukodystrophy. Neurology November 1989; 39: 1415-1422).

COMMENT. X-linked adrenoleukodystrophy is an inborn error of metabolism characterized by adrenal insufficiency and progressive demyelinization. ALD is usually fatal within several years after onset of neurologic abnormalities. A clinically milder form of ALD, adrenomyeloneuropathy, has a later age of onset and slower progression. The authors consider that the dietary erucic acid therapy may be more useful in patients with this milder form of ALD and in presymptomatic boys who express the biochemical defect of ALD.

PEDIATRIC NEUROLOGY BRIEFS (ISSN 1043-3155) ©1989 covers selected articles from the world literature and is published monthly. Subscription requests ($28 US or £15 UK annually; add $5 (£3) for airmail outside North America) may be sent to: Pediatric Neurology Briefs - J. Gordon Millichap, M.D., F.R.C.P. - Editor, P.O. Box 11931, Chicago, IL 60611, USA, or Nat Wst Bnk, 94 Kensington High Street, London W8, UK. The Editor is Professor of Neurology and Pediatrics at Northwestern University Medical School, Chicago, and is presently at Southern Illinois University School of Medicine, Springfield, Illinois, USA.

The possibility of adrenomyeloneuropathy should be considered in any boy with Addison's disease. (Sadeghi-Nejad A, Senior B. N Engl J Med 1990; 322:13-16). These authors measured the plasma concentrations of very long chain saturated fatty acids in eight patients with adrenal insufficiency; five had elevated plasma hexacosanoic acid confirming the presence of adrenomyeloneuropathy. MRI showed brain involvement in all five patients. It was concluded that adrenomyeloneuropathy may present as Addison's disease in childhood.

In a further recent study from the Departments of Pediatric Endocrinology and Radiology, Hopital Saint Vincent de Paul, Paris, France, the MRI detected white matter lesions in two of seven patients with biochemically proven ALD but without neurologic manifestations. The ages at the time of MRI diagnosis were 14 and 21 years. (Auborg P et al. Neurology December 1989; 39:1619-1621). Six of the seven neurologically asymptomatic ALD patients in this study had adrenal insufficiency.

BIOGENIC AMINES IN RETT SYNDROME

The biogenic amines, dopamine, serotonin, and noradrenaline, and their metabolites, were measured in selected brain regions obtained at postmortem from four patients ages 12-30 years with Rett syndrome and are reported from the Departments of Pediatrics, Psychiatry and Neurochemistry, Goteborg University, Goteborg, Sweden. The cause of death was sudden and unexpected in one, severe pneumonitis and pulmonary abscess in one, in association with an operation for scoliosis at 12 years of age in one, and was unrecorded in one. Three of the patients had epilepsy; two were receiving carbamazepine and one sodium valproate at the time of death. Compared to determinations in two adults who had drowned in ice cold water and one killed in a traffic accident, the two older patients with Rett syndrome showed a 50% or greater reduction in biogenic amines in the substantia nigra whereas the youngest patient showed normal or nearly normal levels of biogenic amines in the substantia nigra. The levels were normal in the caudate nucleus, putamen and globus pallidus. The oldest patients had rigidity and dystonic posturing at the time of death whereas the younger 12 year old child was motor disabled secondary to weakness and wasting. The biogenic amine data reflect the clinical patterns of the patients and parallel the neuropathologic finding of reduced melanin content in the neurons of the substantia nigra. (Lekman A, Witt-Engerstrom I, Hagberg BA, Percy AK et al. Rett syndrome: Biogenic amines and metabolites in postmortem brain. Pediatr Neurol Nov-Dec 1989; 5:357-62). Dr. Percy is at the Department of Pediatrics, Baylor College of Medicine, Houston, TX.

COMMENT. Hagberg et al have previously reported a postmortem analysis of brain biogenic amines in an 11 year old Rett syndrome patient in whom the dopamine was markedly reduced in all regions of the brain except the cerebellum and parietal