inheritance; 3) loss of tendon reflexes in lower limbs; 4) dysarthria; and 5) posterior column signs. The more benign 'Acadian' subtype (Barbeau et al, 1984) has previously been differentiated from the classical, French and French Canadian, type of FA and has been attributed to a mutation at the same locus (Keats BJ, Chamberlain S et al. Am J Med Genet 1989;33:266).

LORENZO OIL THERAPY FOR ADRENOLEUKODYSTROPHY

Dietary therapy with glycerol trioleate and glycerol trierucate (Lorenzo oil) was tested in 108 adult patients with adrenomyeloneuropathy phenotype of adrenoleukodystrophy (ALD) at Johns Hopkins Hospital and the Kennedy Krieger Institute, Baltimore, MD. Pattern-reversal visual evoked potentials were used to evaluate visual pathways before and after treatment for 1 year. Very-long-chain fatty acid (VLCFA) levels were markedly reduced, but visual evoked potentials remained abnormal or became abnormal. No patients improved, and there was no evidence that reduction in VLCFA levels benefited or retarded demyelination of visual pathways. (Kaplan PW, Moser HW et al. Visual evoked potentials in adrenoleukodystrophy: a trial with glycerol trioleate and Lorenzo oil. Ann Neurol Aug 1993;34:169-174). (Respond: Dr Kaplan, Department of Neurology, Francis Scott Key Medical Center, 4940 Eastern Avenue, Baltimore, MD 21224).

COMMENT. Moser, in an editorial (Ann Neurol Aug 1993;34:121-122), reviews the development of therapies for adrenoleukodystrophy and describes Lorenzo oil as "a prematurely amplified hope." Trials of the oil in Europe and the USA have failed to demonstrate a significant effect on the rate of progression of the childhood cerebral form of ALD. A possible preventive effect of Lorenzo oil in patients who have not yet developed the neurological disability is under investigation. Of 61 asymptomatic patients treated for a few months up to 4 years at the Kennedy Krieger Institute, one has developed childhood cerebral ALD and 7 have shown progressive demyelination on the MRI. Longer follow-up is required to determine the outcome in the 53 who remain asymptomatic. A depression in platelets was an unexpected side effect of Lorenzo oil therapy.

METACHROMATIC LEUKODYSTROPHY VARIANTS

Clinical, pathological, imaging, and genetic findings in a family with multiple allelic mutations of metachromatic leukodystrophy (MLD) are reported from McGill University, Montreal, and McMaster University, Hamilton, Canada. The propositus, a 23-year-old man, presented at age 18 with a 3-year history of clumsiness and stiffness of gait. Two maternal uncles, ages 48 and 56, were also neurologically impaired. Two siblings, a brother aged 30

and sister aged 25, the mother, and father were clinically normal and had normal electrical studies and MRIs, but their arylsulfatase activity was reduced. Allele-specific amplification confirmed the pseudodeficiency (PD) allele, with no clinical dysfunction, in the mother and her 2 unaffected children. The mother was a PD/MLD compound heterozygote since she was an obligate MLD carrier. The father was a normal (N/MLD) heterozygote. The family was unique in the unusual occurrence of MLD in consecutive generations in the absence of consanguinity. (Francis GS et al. Metachromatic leukodystrophy: multiple nonfunctional and pseudodeficiency alleles in a pedigree: problems with diagnosis and counseling. Ann Neurol Aug 1993;34:212-218). (Respond: Dr Francis, Department of Neurology, Montreal Neurologic Institute, McGill University, 3801 University Street, Montreal, Quebec, Canada H3A 2B4).

COMMENT. Three phenotypes of MLD are recognized based on age of onset: late infantile (0-2 years), juvenile (3-16 years), and adult. The time of onset is determined by the amount of residual arylsulfatase A activity, those retaining some activity having a delayed onset of the disease. A variety of allelic mutations has been identified, including a pseudodeficiency allele with no clinical dysfunction. Cloning of the enzyme gene and genotype identification at the molecular level for the PD and MLD mutations allow appropriate genetic counselling for the families. With these advances in diagnosis, the authors conclude that unnecessary testing of spouses and prenatal screening of pregnancies can be avoided.

MRI NEUROANATOMY OF RETT SYNDROME

The neuroanatomy of 11 females with Rett syndrome (RS) and 15 control subjects investigated in vivo with quantitative MRI is reported from Johns Hopkins University, Baltimore, MD. Compared to age- and gender-matched controls, RS patients had 1) significantly reduced cerebral volume; 2) reduction in brain tissue volumes, especially affecting gray matter; 3) regional variation in cortical gray matter, frontal regions showing the largest decrease; 4) volume reduction of caudate nucleus and midbrain; and 5) increased CSF volume. There was no evidence of an ongoing degenerative process in these patients. (Reiss AL et al. Neuroanatomy of Rett syndrome: a volumetric imaging study. Ann Neurol Aug 1993;34:227-234). (Respond: Dr Reiss, Behavioral Genetics and Neuroimaging, Kennedy Kreiger Institute, 707 North Broadway, Baltimore, MD 21205).

COMMENT. These findings are consistent with one previous neuropathological report, whereas cerebellar pathology was emphasized in another autopsy report of 5 patients (Oldfors A et al. Pediatr Neurol 1990;6:310). See Progress in Pediatric Neurology , PNB Publ, 1991.