distinct subtype of the disease with unique molecular-genetic characteristics. Hemangioblastoma developed in 17% of carriers. (Neumann HPH et al. Pheochromocytomas, multiple endocrine neoplasia type 2, and von Hippel-Lindau disease. N Engl J Med Nov 18 1993;329:1531-8). (Reprints: Dr Neumann, Division of Nephrology and Hypertension, University of Freiburg, Hugstetterstr. 55, D-79106 Freiburg im Breisgau, Germany).

COMMENT. All patients with pheochromocytomas should be screened for von Hippel-Lindau disease by ophthalmoscopy and MRI of the brain. Retinal angiomas are usually asymptomatic. Hemangioblastoma of the CNS has an excellent prognosis if recognized and removed early.

A three-decade investigation of familial pheochromocytoma involving 619 descendants of 3 siblings of German origin, reported from the University of Pittsburgh (Tisherman SE. Arch Intern Med Nov 22 1993;153:2550-6), showed that education and screening decreased mortality. Symptoms on presentation were the classic triad of sweating, nervousness, and headaches. Weight loss, nausea, vomiting, and abdominal pain were more common in children than in adults with pheochromocytoma. Six had von Hippel-Lindau disease manifested by cerebellar hemangioblastoma or retinal angioma, one becoming blind from bleeding. Regular screening and early diagnosis are important to prevent blindness.

METABOLIC AND DEGENERATIVE DISORDERS

VITAMIN E DEFICIENCY FAMILIAL ATAXIC SYNDROME

A total of 8 members of two consanguineous Tunisian families affected with Friedreich's ataxia (FA) phenotype not linked to chromosome 9 were found to have very low levels of serum vitamin E (0.5 mcg/ml cf 8 mcg/ml in controls) in a study at the Institut National de Neurologie, Tunis, and at Centers in Cyprus and France. All patients had typical signs of FA: severe ataxia, bilateral Babinski signs, pes cavus, scoliosis, and absent tendon reflexes and proprioception. The mean age at onset was 11 years, and 6 patients were wheelchair-bound at examination at 20 - 30 years of age. Cardiomyopathy was found in 5. Parents and healthy sibs had normal serum vitamin E levels, pointing to an autosomal recessive trait in affected members. Cystic fibrosis, abetalipoproteinemia, and fat malabsorption syndromes were excluded as possible causes of vitamin E deficiency. This FA with selective vitamin E deficiency was mapped to chromosome 8q, confirming its specificity. (Hamida MB et al. Friedreich's ataxia phenotype not linked to chromosome 9 and associated with selective autosomal recessive vitamin E deficiency in two inbred Tunisian families. Neurology Nov 1993;43:2179-2183). (Reprints: Prof Mongi Ben Hamida, Institut National de Neurologie, La Rabta 1007 Tunis, Tunisia).

COMMENT. The neurological syndrome of vitamin E deficiency is reviewed in an editorial by Dr HJ Kayden, Dept of Medicine, New York University Medical Center, New York (Neurology Nov 1993;43:2167-2169). Abetalipoproteinemia and cholestatic liver disease are characterized by fat malabsorption, resulting in steatorrhea and vitamin E deficiency. In familial isolated vitamin E deficiency, lipid and vitamin E absorption and plasma lipoproteins are normal but conservation of plasma a-tocopherol is poor due to impaired secretion in very low-density lipoproteins. Patients must maintain continuous supplementation with vitamin E (600 IU twice daily, 5 - 10 mg/kg).

The observation that some patients with typical signs of Friedreich's ataxia have a familial vitamin E deficiency syndrome emphasizes the recommendation that all patients with ataxia and neuropathy should have plasma vitamin E determinations. Early supplementation with vitamin E should halt progression of ataxia, and genetic linkage analysis may establish the diagnosis. Asymptomatic sibs should be tested in early childhood, since vitamin E may prevent the onset of ataxia in those affected.

CHOROIDO-CEREBRAL CALCIFICATION WITH RETARDATION

A family of three children with mental retardation, calcification of the choroid plexus, and increased CSF protein is reported from the Riyadh Armed Forces Hospital, and Maternity and Children's Hospital, Riyadh, Saudi Arabia. A 7-year-old Saudi girl born to nonconsanguinous parents showed delayed development. She sat at 3 years, walked at 5, and spoke two- to three-word phrases at 7 years of age. An 8-year-old brother and 5-year-old-sister were similarly affected. Skull radiographs, CT, and MRI showed calcifications along the distribution of choroid plexuses of the lateral ventricles, and calcification involving the caudate nucleus and septum pellucidum. In one child, these findings at 1 year were unchanged over the next 7 years. CSF protein was 180 mg/dl, while phosphate levels in the CSF were low, with a CSF/serum ratio of 0.25 cf normal of 0.73. (Singh B et al. Choroido-cerebral calcification syndrome with retardation. Neurology Nov 1993;43:2387-2389). (Reprints: Dr B Singh, Div Ped Neurology, Southwestern Medical School, 5323 Harry Hines Blvd, Dallas, TX 75235).

COMMENT. Two previous reports of a family of 6 children with a similar syndrome are cited. An autopsy on one patient had shown multiple small subcortical heterotopias and adrenal atrophy (Lott IT et al. Familial amentia, unusual ventricular calcifications, and increased cerebrospinal fluid protein. Neurology 1979;29:1571). That phosphate metabolism may be involved in the pathogenesis is suggested by Singh and co-authors.