CNS LESIONS IN VON HIPPEL-LINDAU SYNDROME

Ninety-seven gene carriers of von Hippel-Lindau syndrome (HLS) were studied at the University of Freiburg, Germany, to evaluate the prevalence and location of CNS lesions. Hemangioblastomas of the CNS were found in 43 patients (44%). Of a total of 93 hemangioblastomas found in 43 patients (44%), 74% were intracranial and 26% were in the spinal cord. Seventy-five percent were cystic and 25% solid. Multiple lesions occurred in 42% of HLS hemangioblastomas but in none of 51 CNS hemangioblastomas without HLS. Cerebella hemangioblastomas were the major cause of death in 82% of the patients with HLS (Neumann HPH et al. Central nervous system lesions in von Hippel-Lindau syndrome. J Neurol Neurosurg Psychiatry Oct 1992; 55:898-901). (Correspondence: Dr. Neumann, Medizinische Klinik, Hugstetter Str 55, D-7800 Freiburg, Germany.)

COMMENT. Other manifestations of von Hippel-Lindau syndrome were as follows: retinal angiomatosis - 44%, renal cysts - 47%, pheochromocytoma - 5%, pancreatic cysts - 30%, and epididymal cystadenoma - 15%. The MRI and gadolinium-enhanced MRI have improved the detection of small CNS lesions which are the initial manifestations of the syndrome and the largest percentage of patients. Multifocal tumor development and recurrence are serious problems in management and prognosis.

METABOLIC DISORDERS

PROPIONIC ACIDEMIA: PROGNOSIS

The neurologic outcome of 20 patients with propionic acidemia was evaluated at the Medical Unit, Institute of Child Health, London, England. In 11 patients who presented in the first week of life, the death rate was high and all were mentally retarded (IQ less than 60), and 3 had mild chorea or dystonia. Of 9 patients with onset after the neonatal period, 4 had a severe movement disorder that evolved following an episode of metabolic derangement. In the late onset group, CT disclosed transient basal ganglia lucencies after episodes of metabolic decompensation. CSF neurotransmitter metabolites were unchanged (Surtees RAH et al. Neurologic outcomes of propionic acidemia. Pediatr Neurol Sept/Oct 1992; 8:333-337). (Communications: Dr. Surtees, Medical Unit, Institute of Child Health, 30 Guilford Street, London WC1N 1EH, England.)

COMMENT. A biotin-responsive propionic acidemia in a newborn is reported from Buenos Aires, Argentina and Paris, France (Cayssials AE et al. Pediatr Neurol Sept/Oct 1992; 8:409 (abstract)). The infant presented at the 4th day of life with partial feeding rejection, drowsiness, tachypnea, hypothermia and hepatomegaly. He developed generalized tonic-clonic seizures and required mechanical respiratory support. Enzyme studies in fibroblasts showed a profound deficiency of propionyl-CoA carboxylase activity and normal values for pyruvate carboxylase. Lab results showed severe metabolic acidosis and