LHERMITTE-DUCLOS-COWDEN DISEASE

Two unrelated patients with clinical characteristics of both Lhermitte-Duclos disease and the autosomal dominant Cowden disease are reported from the Department of Neurology, University Hospital, Leiden, The Netherlands. Patient 1 was a mentally retarded woman with a large head, a high arched palate, dysarthria, mild cerebellar signs, and a single seizure at the age of 37 years. She also exhibited multiple papules on her nose, cheek, forehead and ears and on the oral mucosa. CT scan showed a nonenhancing mass in the cerebellar vermis and left cerebellar hemisphere, enlarged third and lateral ventricles, and a hypodense lesion in the centrum semiovale of the left cerebral hemisphere. A dysplastic gangliocytoma of the left cerebellar hemisphere was diagnosed at surgical resection of the tumor. Microscopic examination of skin biopsies showed trichilemmomas. Bilateral nonmalignant breast tumors were diagnosed at follow-up. The second patient had a history of tonic-clonic seizures in childhood, thyroid surgery at age 38 and he presented with cerebellar signs at 48 years of age. A dysplastic gangliocytoma was removed from the left cerebellar hemisphere. Skin lesions included papules on the face, limbs, and on the gingiva and tongue. Siblings of both patients showed the mucocutaneous lesions, thyroid disease, breast tumors, ovarian tumors, and also neurological signs including macrocephaly, mental retardation, seizures, tremor, and dysdiadochokinesia. The authors propose that Lhermitte-Duclos disease and Cowden disease are manifestations of a single phakomatosis. (Padberg GW et al. Lhermitte-Duclos disease and Cowden disease: A single phakomatosis. Ann Neurol May 1991; 29:517-523).

> COMMENT. Lhermitte-Duclos disease (dysplastic gangliocytoma of the cerebellum) may present in infancy or childhood but the onset of symptoms is more usually delayed until adulthood when a slowly progressive cerebellar syndrome with hydrocephalus is a frequent complication. The characteristic skin lesions of Cowden disease are multiple facial trichilemmomas; hamartomas of other organs, including breast and ovary, occur frequently. Macrocephaly may be an early finding, and the present report suggests that neurological complications of Cowden disease may be under-reported.

VON HIPPEL-LINDAU DISEASE: FAMILIAL SYMPTOM CLUSTERS

The clinical manifestations and prevalence of von Hippel-Lindau syndrome (HLS) were reported from the Department of Medicine, Albert-Ludwigs-Universitat, Freiburg im Breisgau, Germany, and Department of Pathology, University of Zurich, Switzerland. A striking familial clustering of HLS lesions was discovered: angiomatosis retinae in 51% (47/92), haemangioblastoma of the CNS in 46% (42/92), renal lesions in 33% (30/92) and phaeochromocytoma in 24% (22/92). Of the 92 patients, 52 had 1, 31 had 2, and 9 had 3 of the 4 HLS lesions. Of 29 evaluated kindreds, 18 had renal lesions. Some features of HLS were frequently combined whereas others seemed to be almost exclusive. The patterns of lesions within individual kindreds were stable.