Dysplastic gangliocytoma (Lhermitte-Duclos disease) of the cerebellum

Case report

RICHARD W. LEECH, M.D., LEE A. CHRISTOFERSON, M.D., AND ROGER L. GILBERTSON, M.D.

Department of Neuroscience, University of North Dakota and The Neuropsychiatric Institute, Fargo, North Dakota

A case of dysplastic gangliocytoma, or Lhermitte-Duclos disease, of the cerebellum is reported. The patient was the seventh reported survivor of this rare disease. A review of the known biology of the disease allows some optimism. The treatment of choice appears to be surgical resection only.

KEY WORDS - dysplastic gangliocytoma - diffuse ganglioneuroma - Lhermitte-Duclos disease - granule cell hypertrophy - Purkinjeoma

Lhermitte-Duclos disease, or dysplastic gangliocytoma, of the cerebellum is an unusual disorder combining both malformational and neoplastic characteristics in one entity.\(^1,3,8,16\) The most recent and informative review of this rare disease included only 36 cases.\(^1\) We recently had experience with a young man who survived the postoperative period in excellent condition. This case emphasizes the need for greater awareness of this disease and the importance of fundamental knowledge of its biological behavior and morbidity.

Case Report

This 19-year-old ambidextrous man was admitted to the hospital on September 10, 1975, with a 1-month history of progressive, intermittent headaches, not associated with nausea or vomiting. He had been treated unsuccessfully by his father with chiropractic manipulations. During the preceding month he had noticed an increasing drowsiness.

Neurological examination revealed an exophoria with visual acuity of 20-20 on the right and 20-50 on the left. There was bilateral papilledema of five diopters. He exhibited ataxia of tandem gait with ataxia of the upper extremities, the right greater than the left. No nystagmus was noted. The deep tendon reflexes were slightly hyperactive in both upper and lower extremities. There was a suspicious left Babinski sign. Plain x-ray films of the skull showed an abnormally large head with definite ballooning of the right occipital bone (Fig. 1). Routine laboratory studies were within normal limits. A radioactive technetium brain scan appeared normal.
FIG. 1. Skull film shows ballooning of the posterior fossa and thinning of the occipital bone.

On September 11, the patient became semistuporous and complained of intolerable headache. He kept his head in a markedly forward-flexed position and he kept his back in a somewhat arched position. Right ventricular tap was carried out. The intraventricular pressure of the ventricular fluid was over the top of the manometer. After 6 cc of fluid had been removed, the pressure had reduced to 50 cm, and the remaining pressure was reduced gradually by slowly removing 20 cc of clear ventricular fluid. The ventricular needle was left in place and a retrograde four-vessel angiogram was carried out. This showed a large right cerebellar mass with dilatation of the lateral ventricles consistent with obstructive hydrocephalus. A Conray ventriculogram was then performed, and revealed dilatation of both lateral and third ventricles. The proximal one-third of the aqueduct was also dilated and there was no filling beyond this point. The patient was kept on ventricular drainage throughout the remainder of that day, and the next day a suboccipital craniectomy was carried out.

Operation. The cerebellar folia appeared wide, loose, and purplish-gray in color, and this appearance was seen throughout most of the right cerebellum and extending into the right cerebellar tonsil, which was markedly herniated to the level of the upper portion of the lamina of C-2. Large portions of the right cerebellum were removed superiorly and laterally, and repeated frozen sections were obtained without tumor identification. The right cerebellar tonsil was removed along with most of the superior and lateral aspect of the right cerebellar lobe down to the cerebellar nuclei. The cerebellopontine angle was carefully explored, and no evidence of tumor was found. After the area had been thoroughly decompressed, in spite of the lack of definitive tissue diagnosis, the wound was closed and the patient was left on external ventricular drainage through the cannula in the anterior horn of the right lateral ventricle. The correct diagnosis was apparent the following day when appropriately oriented sections revealed that the normal cerebellar cortex was composed entirely of large neurons (Fig. 2).

Postoperative Course. Postoperatively, the patient awakened promptly and was alert. A large amount of slightly bloody cerebrospinal fluid drained from the ventricle and between 300 to 400 cc ventricular fluid continued to drain each 24 hours for 2 weeks. On September 30, the ventricular drainage was terminated; 24 hours later the ventricular pressure was 12 cm H2O, and the ventricular cannula was removed. The patient continued to improve. He was placed on a program of rehabilitation and by October 15 he could care for himself, although he was still somewhat unsteady in walking. By October 21 he could walk without support and was dismissed from the hospital.

Follow-up examination in December, 1975, revealed that the patient had returned to college and was doing well. He stated that he had noticed incoordination of his right extremities, and that, in fact, he had been aware of this for a period of 2 years, although he had not given that information on the admitting history. Further information obtained postoperatively included the fact that he had always had a large head, circumference 62 cm, and had experienced difficulty in purchasing hats. The mother's head circumference was 58 cm.

When last seen in November, 1976, the patient was doing well.

Discussion

Lhermitte-Duclos disease is characterized by the progressive hypertrophy of many cerebellar folia. This hypertrophy may be multifocal, involve one or both hemispheres superiorly or inferiorly, and be deep or superficial. Although it has been considered a hamartoma or neoplasm in the past, microscopically there is a heavily myelinated
Gangliocytoma of cerebellum

Fig. 2. Left: Diffuse layer of large pale neurons replaces normal granule cell layer. H & E, × 10. Right: Higher magnification showing nature of large neurons. H & E, × 64.

molecular layer, absence of normal Purkinje cells, and a deeper layer of large neurons or hypertrophic granule cells (Fig. 2). The general configuration of the cortex is retained, but there is a reduction of white matter. Associated abnormalities include megalencephaly, in about one-half of the cases, leontiasis ossea, polydactyly, hydromyelia, multiple hemangiomata, and heterotopia.

Clinically, the patient usually presents as an adult with a posterior fossa tumor and a history of long-standing vague or poorly delineated mental or neurological abnormalities; however, occurrences in the second decade have been reported. The symptoms have ranged in duration from 3 months to many years, with 1 to 3 years being most common. Some cases have gone unrecognized and others have resulted in sudden unexplained death. Although neither its pathogenesis nor nature are known, we would consider the disease a slowly evolving malformation with little or no neoplastic potential.

It has been suggested that, because of the possible family history and the associated malformations, this condition might be recognized preoperatively. However, even in retrospect there is little information in this case to suggest that the diagnosis could have been made preoperatively. The patient's large head and the skull films suggesting a long-standing chronic problem were the only two positive findings; the family history was negative and there were no associated abnormalities, either physically or mentally.

In spite of the apparent "benign" or slowly developing nature of the disease, this patient was only the seventh case to survive. Such a situation is remarkable in view of the several cases with long asymptomatic periods before diagnosis, several with symptomatic periods exceeding 3 years. The patient of Christian was considered to have a 30-year period of symptoms, but only in the last 10 years were they definite. On the other hand, of the recorded survivors, only one had lived to months at the time of publication. The reason for this paradoxical situation is purely conjectural. All but one of the survivors have been reported since 1955, suggesting that factors such as diagnostic techniques, and surgical and postoperative care may have played a role. The very slow evolution of the process.

J. Neurosurg. / Volume 47 / October, 1977
may allow the patient to compensate to a marked degree, only to have decompensation occur rapidly with onset of symptoms.1,8,12 Some patients have died suddenly postoperatively or following evaluation.4,9,12

With one exception,2 the mode of therapy for each survivor was surgical resection only. In view of the slow progressive nature of the disease and the absence of any adequate evidence for neoplasia, we would suggest that surgical resection remain the treatment of choice until a more rational approach can be derived, and that some optimism is warranted.

References


Address reprint requests to: Richard W. Leech, M.D., Department of Neuroscience, The Neuropsychiatric Institute, 700 First Avenue South, Fargo, North Dakota 58102.