Lhermitte Duclos disease: a rare cause of posterior fossa mass.

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CE - LETTER TO THE EDITOR

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A 57-year-old woman with a history of hypothyroidism, hysterectomy and depression presented to the emergency department (ED) with 3 months of progressive vertigo, ataxia and posterior headache. The patient denied fever, weight loss, vision difficulty or trauma. The patient was directed to the ED because of an abnormal outpatient MRI done earlier that day obtained by her family physician. The patient neither smoked nor drank alcohol. Her medications included: paroxetine, a multivitamin and thyroxine. Her family history was unremarkable.

Physical examination revealed a well-developed woman in no apparent distress, alert and oriented. Her vital signs were normal, and physical examination was remarkable for: lateral nystagmus, dysdiadokokinesis and ataxia. Laboratory studies obtained in the ED including complete blood count, electrolytes and renal function were normal, and neurosurgical consultation was obtained. CT scan demonstrated a left cerebellar mass with coarse calcifications and mass effect on the fourth ventricle, with dilatation of the third and lateral ventricles. MRI demonstrated a large, minimally enhancing cerebellar mass with a striated appearance and confirmed the presence of hydrocephalus (Fig. 1). The diagnosis of Lhermitte Duclos disease (LDD) was made, and the patient was admitted to the hospital. Craniotomy was performed on hospital day three with partial mass resection and external ventricular drain placement. Pathology revealed fully developed gangliocytic transformation of the cerebellar granular and Purkinje cell layers consistent with LDD. Post-operatively, a PET scan demonstrated a residual cerebellar mass with high level of 18-fluoro-deoxyglucose (18-FDG) accumulation consistent with LDD. The post-operative course was uneventful with normal intracranial pressure measurements, and the patient was discharged on hospital day 12 with residual bilateral dysmetria but appropriate functional status. She was maintained on an oral dexamethasone taper with outpatient neurosurgical, occupational and physical therapy follow-up.

First described in the 1920s by Drs. Lhermitte and Duclos, cerebellar gangliocyctoma is an uncommon benign overgrowth of the cerebellar cortex [1]. Cowden's syndrome (CS) is diagnosed in approximately 40% of cases of LDD. CS is an autosomal dominant disorder manifested by multiple hamartomas-neoplasias involving any of the following tissues: breast, uterus, thyroid, skin, genitourinary or colon [2]. Aberrant expression of the tumor suppressor gene, PTEN, may play a role in the development of both LDD and CS. Clinical presentation of LDD is generally in the third or fourth decade without sex predilection and is one of a progressively enlarging posterior fossa mass with mass effect and cerebellar signs. Diagnosis can be made pre-operatively with MRI [1]. Pathology is confirmatory. Functional neuroimaging such as 18-FDG PET scan can further aid in diagnosis, revealing increased uptake due to hypervascularity [3]. Treatment includes surgical resection and relief of elevated intracranial pressure. Although there have been no reports of malignant transformation of LDD,

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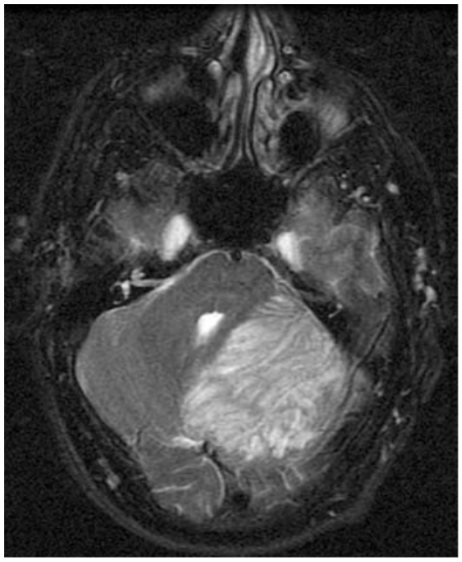

Fig. 1 Axial FSE T2 image through the posterior fossa demonstrates a well-circumscribed T2 hyperintense lesion that expands the left cerebellar hemisphere, exerting mass effect on the fourth ventricle. Linear parallel striations isointense to cerebellar grey matter along its surface correspond to enlarged cerebellar folia with dilated gyri

regrowth of gangliocytic tissue may occur [3]. Patients with LDD should be screened for CS due to the increased risk of neoplasia. Female CS patients have lifetime risks of 25–50% and 10% for breast and thyroid cancer, respectively [1]. Vigilant follow-up is required for both conditions [4].

LDD is a highly unusual cause of cerebellar mass and dysfunction, and its association with CS is important due to malignant potential. Both emergency and internal medicine physicians should be cognizant of the clinical manifestations, diagnosis and treatment.

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