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Case report

Intramedullary ependymoma associated with Lhermitte–Duclos disease and Cowden syndrome

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Abstract

The authors describe the case of a 45-year-old man with progressive gait ataxia and sensorimotor deficits of the upper and lower extremities. The patient had been diagnosed earlier with Lhermitte–Duclos disease (LDD) in the left cerebellar hemisphere and Cowden syndrome (CS). MR imaging studies revealed an intraspinal tumor at C6–C7. Microsurgical gross total resection of the tumor was achieved. Histolopathological examination revealed an intramedullary ependymoma. Postoperatively, neurological deficits gradually improved. This is the first reported case of ependymoma in a patient with LDD and CD. Coexistence of an intraspinal ependymoma with cerebellar LDD and CS appears to be rare, but can lead to treatment failure if missed.

Keywords: Lhermitte–Duclos disease; Cowden syndrome; Ependymoma; Spinal tumor

1. Introduction

Lhermitte–Duclos disease (LDD) or dysplastic cerebellar gangliocytoma is a rare slowly progressive cerebellar tumor characterized by an enlargement of cerebellar foliae [1,2]. Its pathogenesis is still unclear [3]. Frequently, LDD is associated with Cowden syndrome (CS) [4–6]. CS is a rare autosomal dominant disorder characterized by multiple hamartomas with malignant features [5,7]. Various additional abnormalities including meningiomas or vascular malformations have also been shown to be associated with CS [8–10]. To our knowledge, no instance of ependymoma in CS has been described thus far. Here, we report the first case of an intraspinal intramedullary ependymoma in an adult patient with LDD and CS.

2. Case report

2.1. History

A 45-year-old man had been diagnosed with LDD five years earlier, when a nonenhancing mass in the left cerebellar hemisphere was shown by MR imaging (Fig. 1). At that time he suffered from headache and dysmetria of the left hand. The patient opted not to undergo surgery. Since he and several other members of his family also had multiple lipomas, hamartomas of the skin, and polyps of the colon a diagnosis of CS was suspected. This was confirmed by PTEN gene molecular analyis, which revealed a mutation on chromosome 10, consisting of a nonsense T to G mutation at nucleotide position 663–672, exon 7 (Fig. 2). The patient was admitted to our department because of slowly progressive spinal ataxia since one and a half years with gait deterioration, as well as mild distal paraparesis and paresthesias in both arms.

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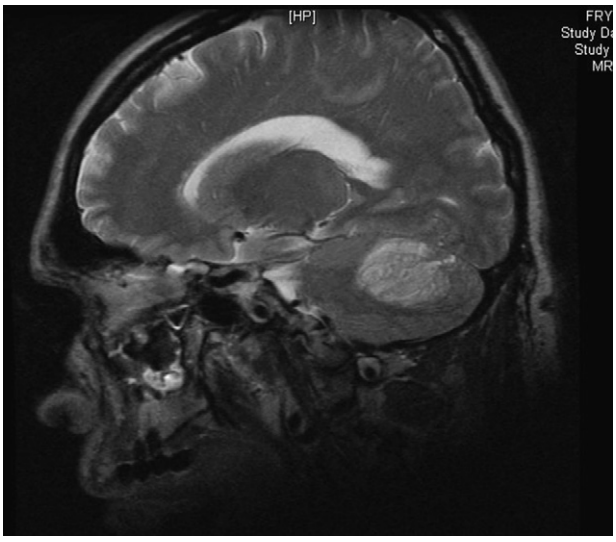

Fig. 1. T2-weighted sagittal MR image scan of the neurocranium demonstrating a nonenhancing hyperintense mass in the left posterior fossa corresponding to the known picture of Lhermitte–Duclos disease without signs of hydrocephalus.

2.2. Examination

Neurological examination revealed weakness of dorsal flexion of both feet (grade 3/5), and symmetric weakness of the quadriceps muscles (grade 4/5). There was also a mild weakness of finger extension (grade 4/5). Furthermore, hypaesthesia of the ulnar forearm was noted. Reflexes were diminished in both upper and lower extremities. Babinski's sign was negative. Dysmetria and dysdiadochokinesia were present on the left. There was pronounced gait ataxia. MR imaging studies of the neurocranium did not show an increase in size or signal alterations of the well-known LDD in the posterior fossa. MR imaging studies of the spine revealed an intramedullary tumor at C6 and C7 with high signal intensity in T1-weighted images after contrast with good demarcation to the normal spinal cord (Fig. 3). T2-images showed pronounced perifocal medullary edema. There were no polar cysts.

2.3. Treatment

Microsurgical gross total resection of the tumor was performed via a C6 and C7 laminectomy. The tumor mass was solid with good demarcation from the adjacent normal cord tissue. The tumor was moderately vascularized, and it could be removed without difficulty. The extent of tumor removal was confirmed by postoperative MR images. The histological examination demonstrated an ependymoma WHO grade II with papillary, epithelial, and mixed cells in typical pseudorosette formations (Fig. 4).

2.4. Postoperative course

The postoperative period was without adverse events. Following surgery, there were no new neurological deficits. Gradual improvement of gait ataxia and paraparesis was observed with physiotherapy. At follow-up nine months postoperatively, neurological deficits were almost normalized.

3. Discussion

LDD or dysplastic cerebellar gangliocytoma is characterized by focal or diffuse enlargement of cerebellar foliae

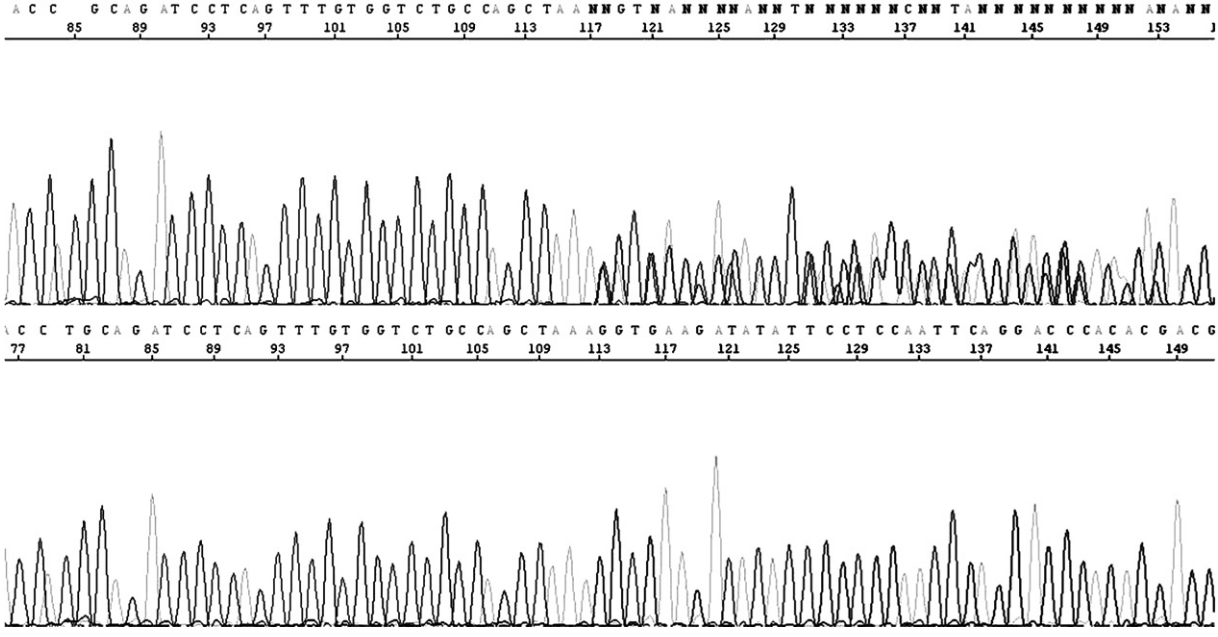

Fig. 2. PTEN_ex7-FOR, Mutation 663_672delGGTGAAGATA in exon 7 of the PTEN gene. Top, mutated sequence in patient; bottom, normal control sequence.