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Case study

Diagnosis and treatment for pure aqueductal tumor

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ABSTRACT

Pure aqueductal tumor (PAT) typically originates from pure aqueductal region and is extremely rare. It is radiographically similar to tectal glioma. We examined two patients with PATs who were diagnosed with pilocytic astrocytoma and rosette-forming glioneuronal tumor. Both cases showed a progressive clinical course. It is important to distinguish between PAT and tectal glioma by radiographic imaging because the treatment strategy is different. While observation is common for tectal gliomas, a biopsy is recommended at the same time of endoscopic third ventriculostomy for PAT with hydrocephalus. Low-grade PATs show an aggressive clinical course in some cases. Our two cases also showed aggressive course in spite of no genetic aggressive mutations. Sagittal view by constructive interference in steady state (CISS) imaging was helpful to make an accurate diagnosis of PAT. Close observation is needed if PAT is diagnosed as low-grade tumor.

1. Introduction

Periaqueductal tumors usually originate from the tectum or the pineal body [1–6]. Pure aqueductal tumor (PAT) is one of periaqueductal tumors that are radiographically similar to tectal glioma. However, PATs typically originate from the pure aqueductal region and are extremely rare. Only 16 cases have been previously reported. Only two cases were diagnosed as pilocytic astrocytoma (PA) and none as rosette-forming glioneuronal tumor (RFGT) [1,7–17]. We examined two aggressive cases of PAT that were diagnosed as PA and RFGT. Genetic analysis were first performed. The treatment strategy for periaqueductal tumors differs depending on their primary origin. If tectal glioma is suspected, observation is recommended [1–3,5]. However, PATs often occupy the aqueductal lumen; therefore, non-communicating hydrocephalus is observed. Surgical resection is performed in some cases via open or endoscopic approach [1,7,9,10–12,14,16,17]. Although it is sometimes difficult to distinguish between tectal glioma and PAT by radiographic imaging, determining the origin of the tumor is important. In addition, the treatment strategy for PATs is also controversial. In some cases, surgical resection is performed, while only biopsy and endoscopic third ventriculostomy (ETV), or even just observation, is preferred in other cases [1,7,9,10–12,14,16,17]. The methods to distinguish PATs from other periaqueductal tumors radiographically and treatment strategies for PATs are discussed in this report.

2. Case reports

Case 1 A 29-year-old male presented with headache, and magnetic resonance imaging (MRI) showed a periaqueductal tumor. The tumor was seen as high-intensity regions on T2-weighted images (T2WI) and low-intensity regions on T1-weighted images (T1WI), with abnormal gadolinium (Gd) enhancement. Tumor origin from tectal region could not be ruled out by only radiographic images. Two years later, the enlarging tumor caused non-communicating hydrocephalus (Fig. 1A). ETV and contemporary biopsy were performed using a flexible scope. The diagnosis of PAT was made by operative view (Fig. 1B). Hydrocephalus was improved; however, a year later after operation the tumor became enlarged again. Tumor removal was performed via the Telovelar approach. The tumor was strongly attached to the aqueduct. Therefore, total removal was difficult. Histopathological diagnosis was RFGT. MIB-1 index was 0.3%. Since the second surgery, the tumor has not shown enlargement. **Case 2** A 19-year-old male with neurofibromatosis type 1 (NF1) presented to another hospital with headache in 7 years ago. MRI showed a small-sized tumor of 13 mm in diameter located within the aqueduct; however, hydrocephalus was not observed (Fig. 2B). He complained of headaches again 3 years later. MRI showed an enlargement of the tumor and slight hydrocephalus. He refused surgery and chose observation. The frequency of headache had increased by further 4 years later. MRI showed further enlargement of the tumor, and hydrocephalus had worsened. The lobulated tumor, 27 mm in diameter, showed slight high intensity on T2WI, low intensity on T1WI, and partial Gd enhancement. Determining the origin of tumor by Gd

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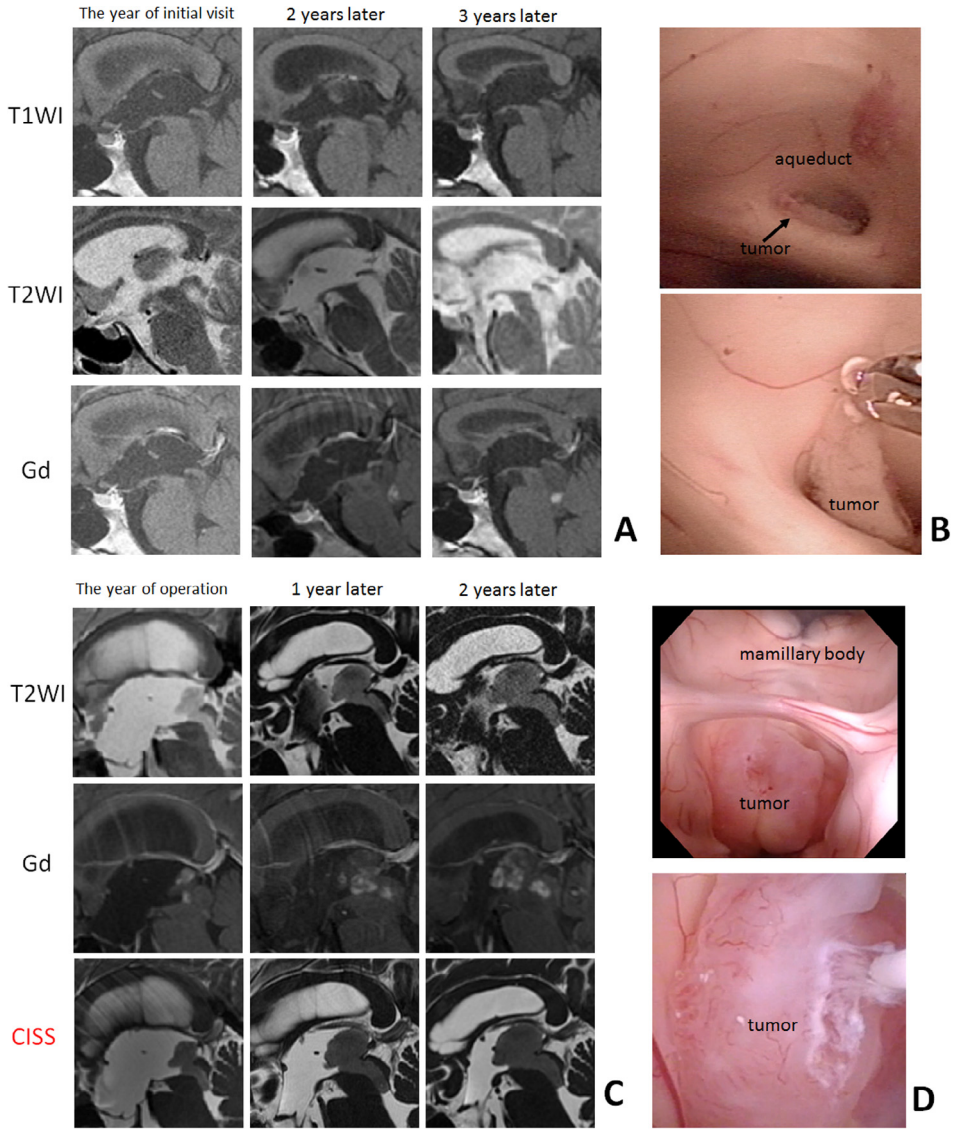

Fig. 1. (A) Time-dependent change in Case 1 showed tumor enlargement. The size of tumor was 7.2 mm × 12 mm in the year of initial visit and 9.2 mm × 25.9 mm 3 years later. The attachment of the tumor was not clear, especially by Gd-enhanced image. T1WI and T2WI were helpful; however, it was difficult to make an accurate diagnosis of PAT by only radiographic imaging. (B) The diagnosis of PAT was intraoperatively made. Adhesion to surrounding tissues was strong. (C) Time-dependent change in Case 2 showed tumor enlargement. The size of the tumor was 12.3 mm × 4 mm in the year of initial visit and 37.9 mm × 28.1 mm 6 years later. The attachment of the tumor was not clear, especially by Gd-enhanced image. The diagnosis of PAT was accurately made by CISS. (D) It was intraoperatively observed that the tumor was attached to the aqueduct and it was hardly bleeding.

enhancement and T2WI imaging was difficult for Case 1. Therefore, a sagittal view of constructive interference in steady state (CISS) was performed in Case 2. That image showed that the tumor did not originate from the tectum or pineal body but independently arose from the aqueduct. Therefore, a diagnosis of PAT was suspected (Fig. 1C). ETV and contemporary biopsy were performed using a flexible scope. A whitish tumor was identified at the aqueduct. MRI 1 day postoperatively showed patency of the floor of third ventricle. The histopathological diagnosis was PA. MIB 1 index was 0.6%. Two years later after operation, he was still asymptomatic; however, the tumor became enlarged to 40 mm.

Genetic analyses No whole chromosomal copy number alterations (CNAs) were detected by comparative genomic hybridization (CGH) in both cases. Mutations of BRAF V600E, and IDH1/2 were analyzed by direct sequencing. IDH1 codon 132, IDH2 codon 172, and BRAF codon 600 were found to possess the wild-type sequence in both cases.

3. Discussion

Tumors in the tectal and pineal regions are defined as periaqueductal tumors. PAT, a rare entity, is a type of periaqueductal tumor. Tectal glioma, often diagnosed as pilocytic astrocytoma, does not normally show a progressive course and generally stops growing after childhood. Therefore, if tectal glioma is suspected by radiographic imaging, observation is appropriate. If the tumor shows enlargement and is causing hydrocephalus, biopsy and ETV are considered [1–3,5]. Only 16 cases of PAT have been reported. Pathologies of PAT include heterotopias, pilocytic astrocytomas, subependymomas, ependymomas, ependymoblastomas, glioneuronal tumors, and medulloblastomas [1,7,9,10–12,14,16,17]. PAT often causes non-communicating hydrocephalus. Therefore, surgical resection, biopsy, and ETV or VP shunt were often considered. Only three of the reported cases had progressive tumor enlargement, while the remaining 13 cases showed no interval changes.