Dysembryoplastic neuroepithelial tumor presenting as a hypothalamic hamartoma in a child with gelastic seizures: case report

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Dysembryoplastic neuroepithelial tumors (DNETs) are benign intracranial tumors of neuroglial origin, mostly located in the supratentorial regions and particularly in the temporal lobe. Few cases of DNETs in the hypothalamus have been described. The authors present the case of a DNET in the hypothalamus. The 5-year-old girl with complaints of limb and gelastic seizures was admitted to the neurosurgical department of Xiangya Hospital. Neurological examination findings were unremarkable. MRI showed isointensity without significant enhancement on T1- and T2-weighted images. The lesion exhibited clearly defined borders on the sagittal, coronal, and axial images. The preliminary diagnosis was hypothalamic hamartoma (HH); however, the lesion was surgically removed, and histopathological examination confirmed the diagnosis of a DNET. Hypothalamic DNETs are extremely rare. Based on their clinical manifestation and imaging, DNETs are easily misdiagnosed as HHs. Diagnoses apart from HHs must be entertained when a hypothalamic lesion is being investigated.

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waves progressively increased. The frequency of slow waves lessened gradually. The seizure ceased when the frequency reached 1–2 Hz.

A noncontrast cerebral CT scan showed a circular lesion (32 Hounsfield units) about 4 mm in diameter in the left area of the suprasellar cistern. The lesion showed no calcification (Fig. 1). MRI showed a lesion in the hypothalamus that had isointensity without significant enhancement on T1-weighted, T2-weighted, and FLAIR images. The lesion had clearly defined borders on the sagittal, coronal, and axial views (Fig. 1). A hypothalamic hamartoma (HH) was the preliminary diagnosis.

Operation and Follow-Up

The lesion was exposed via pterional approach. A gray tumor with dimensions of $1 \times 0.8 \times 0.8$ cm could be seen. The lesion had clearly defined borders and abundant vascularity. Subtotal resection was performed using a microsurgical technique because the posterior portion of the lesion had close adhesion to the pedunculus cerebri and hypothalamus. The patient had an uneventful postoperative recovery. Over the 12-month follow-up period, the patient was neurologically intact and seizure free (Fig. 2).

The results from the histopathological examination confirmed that the lesion was a hypothalamic DNET (Fig. 3). The histopathology showed isolated neurons with large nuclei characteristically floating within the mucoid substance, encircled by a glial component, which was composed mainly of oligodendroglia-like cells. Immunohistochemical staining demonstrated the following findings: negative p53 and isocitrate dehydrogenase 1 expression and positive glial fibrillary acidic protein, oligodendrocyte transcription factor 2, NeuN, and O6-methylguanine-DNA methyltransferase expression.

Discussion

DNETs are benign tumors that belong to neuronal and mixed neuronal-glial tumors. The tumors typically manifest as medically intractable, partial complex seizures in children and adolescents. Consistent with the characteristics of DNETs, our patient was a 5-year-old child who presented with a 4.6-year history of seizures without known precipitating events. DNETs typically occur within the supratentorial cortex and most commonly in the temporal lobe. On MRI, DNETs are characterized by a multinodular structure, rendering a bubbly appearance. Mass effect and perilesional edema are usually absent even in voluminous lesions. A triangular pattern distribution and a FLAIR rim are typical imaging findings that can be observed in cases of DNETs. In the current case, MRI on FLAIR and contrast enhancement imaging had an isointense signal, and the patient presented with gelastic seizures, together rendering the mass diagnostically indistinguishable from an HH. HHS are masses that consist of well-differentiated ectopic neurons interspersed with glial cells, and they are located in the tuber cinereum of the hypothalamus. They are congenital malformations rather
than cancerous tumors. Prototypical clinical presentations of HHs include precocious puberty and gelastic seizures that begin in infancy. The signal intensity of HH lesions on MRI is hypointense on T1-weighted sequences and hyperintense on T2-weighted sequences. Therefore, based on age, clinical manifestations, and imaging, the preoperative diagnosis was HH. Only after performing histopathological examination was the diagnosis of a DNET confirmed. DNETs have intracortical nodules within a microcystic, vacuolated background. The structure additionally contains columnar architecture of uniform oligodendrocyte-like cells, which are lined around variously shaped and sized microcysts. This morphology is called “specific glioneuronal elements,” which, accompanied by numerous interspersed floating neurons, is the most distinctive pathological feature of DNETs. However, not all DNETs have specific glioneuronal elements, and some DNETs exhibit a growth pattern similar to that of diffuse gliomas. Therefore, the differential diagnosis includes gangliogliomas, low-grade diffuse gliomas, focal cortical dysplasia, oligodendrogliomas, pilocytic astrocytomas, and diffuse astrocytomas.

For HH, there are various hypothalamic lesions that need a differential diagnosis. Some lesions originate from the suprasellar cistern or third ventricle, which can invade the hypothalamus, such as craniopharyngiomas, epidermoid cysts, Rathke’s cleft cysts, and colloid cysts. Some lesions are simply located in the hypothalamus, masquerading as an HH. Fukunaga et al. reported a single case with hypothalamic-pituitary germinoma presenting as generalized hypohidrosis. Wang et al. reported a case with exophytic chiasmatic/hypothalamic glioma. The tumor was partially removed by endoscopic transsphenoidal surgery, with good preservation of hypothalamic and endocrine functions. A rosette-forming glioneuronal tumor originating in the hypothalamus and manifesting with precocious puberty has also been reported. Bognár et al. described a case of intracranial osteolipomas that occurred in the region of the tuber cinereum. The patient presented with an ovarian cyst and signs of precocious puberty. A DNET located in the hypothalamus has also been described in a previous case report. That case involved a 50-year-old man with a 9-year history of generalized tonic-clonic sei-

![FIG. 2. The lesion location is marked by the white arrow on axial (A), sagittal (B), and coronal (C) images. As the 12-month follow-up MRI examination shows, most of the tumor tissues have been removed.](image)

![FIG. 3. The most distinctive pathological features of DNETs are their “specific glioneuronal elements” accompanied by numerous interspersed floating neurons. Specific glioneuronal elements are composed of ganglion cells suspended within a basophilic mucoid matrix encircled by oligodendroglia-like cells. H & E, original magnification ×200. Figure is available in color online only.](image)
not match the typical HH, we recommend doing a biopsy before a nonresective intervention.

Complete tumor resection is considered to be a major prognostic factor for DNETs in most studies. However, whether DNETs need lesionectomy only, rather than extended resection, is a matter of debate, especially when the lesion is located within the temporal lobe. Some studies recommend surgical planning based on intraoperative electrocorticography combined with MRI. However, one study reported a case in which intraoperative monitoring with electrocorticography and tailored resection did not improve seizure outcome. In the current case, the tumor was located in an area of the hypothalamus atypical for common DNETs. The lesion was adherent to important surrounding nerve and vessel structures, such as the optic tract, pituitary stalk, and superior hypophyseal artery. The lesion was carefully surgically separated from normal tissue. Subsequently, the lesion was also found to be adherent to the pedunculus cerebri and hypothalamus. To avoid severe postoperative functional impairment and homeostatic disturbances, the lesion was not removed completely. At the 12-month follow-up visit, the patient was seizure free and was not taking antiepileptic drugs. Incomplete resection is usually regarded as the major cause of surgical failure. Some studies suggest that the follow-up should continue for a minimum of 3 years to reliably assess seizure outcomes, as some children may only achieve short-term remission. Therefore, our case requires longer follow-up to fully evaluate the therapeutic effects.

Although DNETs are typically benign, the presence of such tumors can result in refractory epilepsy that interferes with a patient’s daily life. Surgery is the most effective treatment. Patients have experienced favorable outcomes following complete resection. Preoperative assessments are necessary to accurately diagnose the lesion from a range of similarly presenting lesions. Complete resection of the DNET is crucial to achieve a favorable long-term outcome, but severe postoperative complications must be prevented.

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References


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Author Contributions

Conception and design: Cai. Acquisition of data: Cai. Analysis and interpretation of data: Wang. Drafting the article: Cai. Critically revising the article: Yang. Wang. Reviewed submitted version of manuscript: Yang. Wang. Approved the final version of the manuscript on behalf of all authors: Yang.

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