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Case Report

EPITHELIOID GLIOBLASTOMA IN A CHILD WITH TUBEROUS SCLEROSIS

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ABSTRACT

Tuberous sclerosis (TS) is a rare autosomal dominant disease. This case report reports a 3-year-old boy with epitheloid glioblastoma in the TS region. The patient had right-sided hemiparesis and facial nerve palsy and underwent a frontotemporal craniotomy. The patient was treated with a bone graft, a very rare method. Therefore, a yearly MRI of the brain is recommended in TS.

KEYWORDS: *epithelioid glioblastoma, tuberous sclerosis, SEGA, TSC2, neurosurgery*

DISCUSSION

Tuberous sclerosis (TS) complex is a rare autosomal dominant disease. It occurs in 1 in 6,000 individuals (1). TS is associated with the manifestation of (sub)cortical tubers, calcified subependymal nodules and subependymal giant cell astrocytomas (SEGAs) (2). Brain tumors associated with TS are rather unusual, affecting only 6–14% of patients; SEGAs account for the majority (3).

Herewith, we report the case of a 3-year-old girl with epithelioid glioblastoma in the context of TS. Diagnosis was set with the Sanger sequencing method. A c.4846 C>T (p.Gln1616stop) mutation was identified during the neonatal age on the exon of the TSC2 gene. She also had suffered drug resistant epilepsy under triple antiseizure medication (oxcarbazepine, topiramate, and valproic acid).

Two weeks before her admission, she exhibited right sided hemiparesis and ipsilateral central facial nerve palsy. Magnetic Resonance Imaging (MRI) examination of the brain revealed a space occupying lesion frontotemporally on the left, with slight calvarial erosion (Fig.1).

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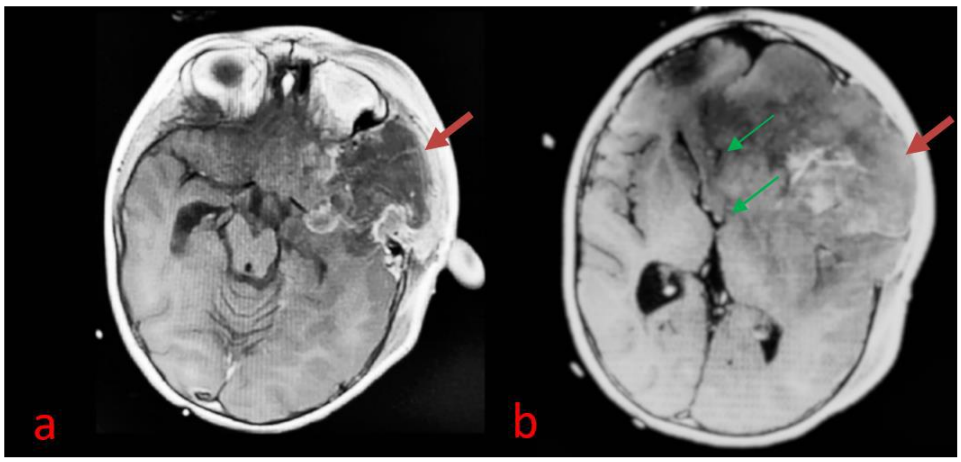

Fig. 1. *a): Space occupying lesion of left temporal pole and orbital gyri with bone erosion, adjacent to the upper orbital wall (red arrow); b): It is extraventricular, extends to inferior frontal gyrus (red arrow), and compresses the left frontal horn (thin green arrows). Histology exhibited epithelioid glioblastoma*.

Initially, she underwent a frontotemporal craniotomy under neuronavigation. The operation ceased due to circulatory instability and massive cerebral edema. Consequently, the bone flap was not placed back. In addition to that, a right-sided ventriculoperitoneal shunt system was implanted one week afterwards due to hydrocephalus. An adjunct second ventral catheter was inserted three weeks later due to persistent meningocele (Fig.2).

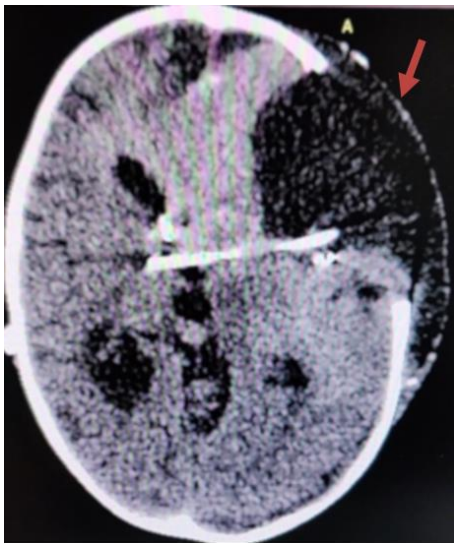

Fig. 2. *Ventriculoperitoneal system with two central catheters due to persistent meningocelcele (red arrow) and deficient communication between the two lateral ventricles.*

Histology revealed epithelioid glioblastoma. The child had undergone radiotherapy with 34.2 Grays total dose and three cycles of Temozolamide. Seven months afterwards, we implanted the custom-made bone graft and resected a new lesion of the left temporal pole, which had been considered local tumor recurrence; this lesion was identified as a SEGA (Fig.3).

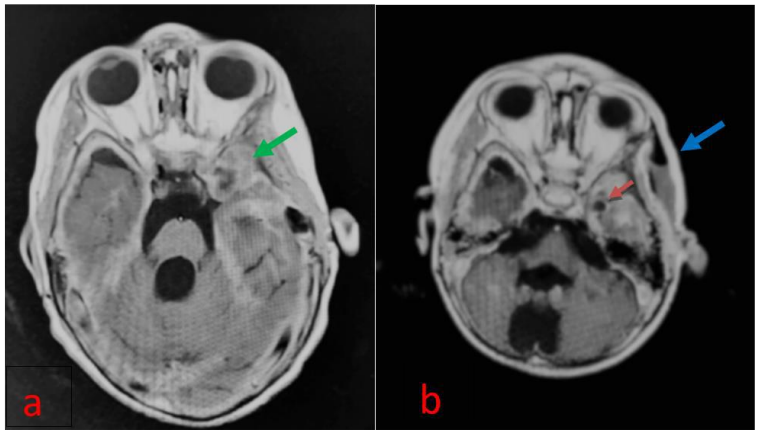

Fig. 3. ***a****): New lesion at the left temporal pole (green arrow) after the fourth surgical procedure;* ***b****): Bone graft implantation (thick blue arrow). The anatomical region of the excised left temporal pole lesion, which was identified as a SEGA (small red arrow).*

So far there are only seven cases of patients with occurrence of glioblastoma in the context of TS. In all of them, glioblastoma did not ensue as a transformation of the SEGA lesion (2). Malignant manifestations of TS have an incidence of 2% and afflict mostly younger patients (3). The association between high malignant tumors and primary lesions is not clear and they probably develop independently.

According to the updated surveillance criteria of TS, MRI of the brain should take place every one to three years (4). We advocate that the annual follow-up control should involve all TS patients and not only ones with large SEGAs.

Conflict of interest

The authors declare that they have no conflict of interest.

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