ABSTRACT

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Effective treatment of a BRAF V600E-mutant epithelioid glioblastoma patient by vemurafenib: a case report.

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Epithelioid glioblastoma (E-GBM) is a recently described variant of glioblastoma (GBM) which is associated with short survival and now added as a provisional entity to WHO 2016 classification of central nervous system tumors. About half of these tumors show the BRAF mutant. Therefore, this is a target of special interest for this group of patients. Meanwhile, unlike conventional glioblastoma, E-GBM lacks specific prognostic markers. We described a case of a long-term surviving 37-years-old men patient diagnosed with a BRAF V600E and TERT mutated E-GBM with wild-type in the isocitrate dehydrogenase gene (IDH wild-type). The tumor displayed atypical exophytic growth, an obvious proliferation of vascular endothelial cells, especially tumor tissue can be seen under subarachnoid space. Notably, tumor tissue was found under subarachnoid space. After postoperative conventional treatment options were exhausted, vemurafenib treatment was initiated. The patient remained clinically stable, and follow-up magnetic resonance images were consistent with stable disease for the following fifteen months up to now. Whole-exome sequencing analysis and RNA-seq results of formalin-fixed and paraffin-embedded tissue revealed nine mutant genes (AHNAK2, BFSP1, BRAF, CNTNAP3, DNHD1, MTOR, NFATC3, NOM1). For E-GBM patients, the use of BRAF inhibitors combined with inhibitors of these seven genes may be a useful remedial treatment option.

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