**Case Reports** 

Neuropathology, 2023 Nov 2. doi: 10.1111/neup.12948. Online ahead of print.

## SEGA-like circumscribed astrocytoma in a non-NF1 patient, harboring molecular profile of GBM. A case report

Seiji Yamada <sup>1</sup> <sup>2</sup>, Motoki Tanikawa <sup>1</sup>, Yuko Matsushita <sup>3</sup>, Ryota Fujinami <sup>1</sup>, Hiroshi Yamada <sup>1</sup>, Kaishi Sakomi <sup>4</sup>, Tomohiro Sakata <sup>1</sup>, Hidehito Inagaki <sup>5</sup>, Hideaki Yokoo <sup>6</sup>, Koichi Ichimura <sup>3</sup>, Mitsuhito Mase <sup>1</sup>

Affiliations

PMID: 37919875 DOI: 10.1111/neup.12948

## **Abstract**

Subependymal giant cell astrocytoma (SEGA) is a low-grade periventricular tumor that is closely associated with tuberous sclerosis complex (TSC). SEGA typically arises during the first two decades of life and rarely arises after the age of 20-25 years. Nevertheless, it has also been reported that glioma histologically resembling SEGA, so-called SEGA-like astrocytoma, can arise in neurofibromatosis type 1 (NF1) patients, even in the elderly. Herein, we report a case of SEGA-like circumscribed astrocytoma arising in the lateral ventricle of a 75-year-old woman. Whole-exome sequencing revealed a somatic variant of NF1. Methylation array analysis led to a diagnosis of "methylation class glioblastoma, IDHwildtype, mesenchymal-type (GBM, MES)" with a high calibrated score (0.99). EGFR amplification, CDKN2A/B homozygous deletion, chromosomal +7/-10 alterations, and TERT promoter mutation, typical molecular abnormalities usually found in GBM, were also observed. While most reported cases of SEGA-like astrocytoma have arisen in NF1 patients, the patient was neither TSC nor NF1. Near total removal was accomplished with endoscopic cylinder surgery. At the 36-month follow-up, there was no tumor recurrence without adjuvant therapies. This clinical behavior did not match GBM. SEGA-like astrocytoma of the elderly is rare, and this is the oldest case reported so far. In addition, high-grade molecular features found in circumscribed tumor remain unclear. Further investigations among larger series are needed for clarifying the underlying molecular mechanisms.

**Keywords:** DNA methylation array; NF1; SEGA; TSC; whole-exome sequencing.

© 2023 Japanese Society of Neuropathology.

PubMed Disclaimer

1 di 1 09/11/2023, 16:30