

ABSTRACT

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Story of the solitary subependymal giant cell astrocytoma: A case report and literature review.

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Subependymal giant cell astrocytoma (SEGA) is the characteristic benign, slow-growing brain tumor seen in tuberous sclerosis (TS). There are several case reports of a diagnosis of SEGA in patients with no clinical or radiological diagnosis of TS. However, there is limited literature describing the tumor genetics in such cases. We report a case of a 17-year-old girl who was diagnosed with SEGA bearing the TSC2 mutation, while testing negative for TSC mutations on germline testing. We also did a literature review of studies that reported the genetics behind solitary SEGAs. Genetic testing of both the tumor itself and germline genetic testing can provide valuable information with clinical implications, for example, the basis for the need of close surveillance in TS patients.

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