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Subependymal giant cell astrocytoma (SEGA), unrelated to tuberous sclerosis, NTRK positive

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A 11-year-old child presented with hydrocephalus-related symptoms. MR demonstrated, in the post T1-weighted image, an enhancing intraventricular mass in the frontal horn of the right lateral ventricle. The patient underwent neurosurgery and histology showed it to be a neoplasm with compact architecture, high cellularity, large cells with eosinophilic cytoplasm, in absence of mitosis and/or vascular proliferation and/or necrosis (fig. 1). Immunohistochemistry revealed positivity for GFAP and S100. The diagnosis was subependymal giant cell astrocytoma (SEGA). SEGA is a rare glial neoplasm typically located in the wall of the lateral ventricles and usually associated with tuberous sclerosis (TS), an autosomal dominant syndrome harbouring mutations in the TSC1 and TSC2 genes, although cases unrelated to TS are reported. Our case fits into this context of rarity: indeed, the patient was referred for genetic counselling after histological diagnosis, but no alteration in tuberous sclerosis-related genes was found. Although the last two editions of the World Health Organization's classification of central nervous system tumours (WHO-CNS2016 and WHO-CNS2021) have introduced real 'revolutions' in the morpho-molecular aspects of most primary brain neoplasms, SEGA has not substantially changed its classification, always maintaining its features (grade 1 according to WHO-CNS2021), and constituting one of the longest-lived entities of all CNS tumours [1]. Probably because of the rarity of SEGA -compared to neoplasms with extremely higher incidence, prevalence and mortality- histologic expression of predictive targets in SEGAs has not been studied to date. Our immunohistochemistry results were: NTRK+ (fig. 2), ALK-, PDL1-, PD1-, CTLA4-. To date, SEGA therapy is limited to m-TOR inhibitors, such as rapamycin [2], and therefore the immunohistochemical NTRK-positivity could potentially broaden the ever-expanding landscape of tumours treatable with TRK-inhibitors (immunohistochemical data, ours, which may be validated by subsequent molecular confirmation in larger case series), whereas our results suggest no correlation with immunocheckpoint expression.

Article information and declarations

Ethics statement

All procedures performed are in accordance with the Helsinki Declaration.

Author contributions

Veronica Parrella – is responsible for drafting the article.

Jacopo Ferro – is responsible for editing the images.

Chiara Trambaiolo Antonelli – is responsible for the bibliography.

Gabriele Gaggero – is responsible for the conception and supervision of the article.

Conflict of interest

None declared

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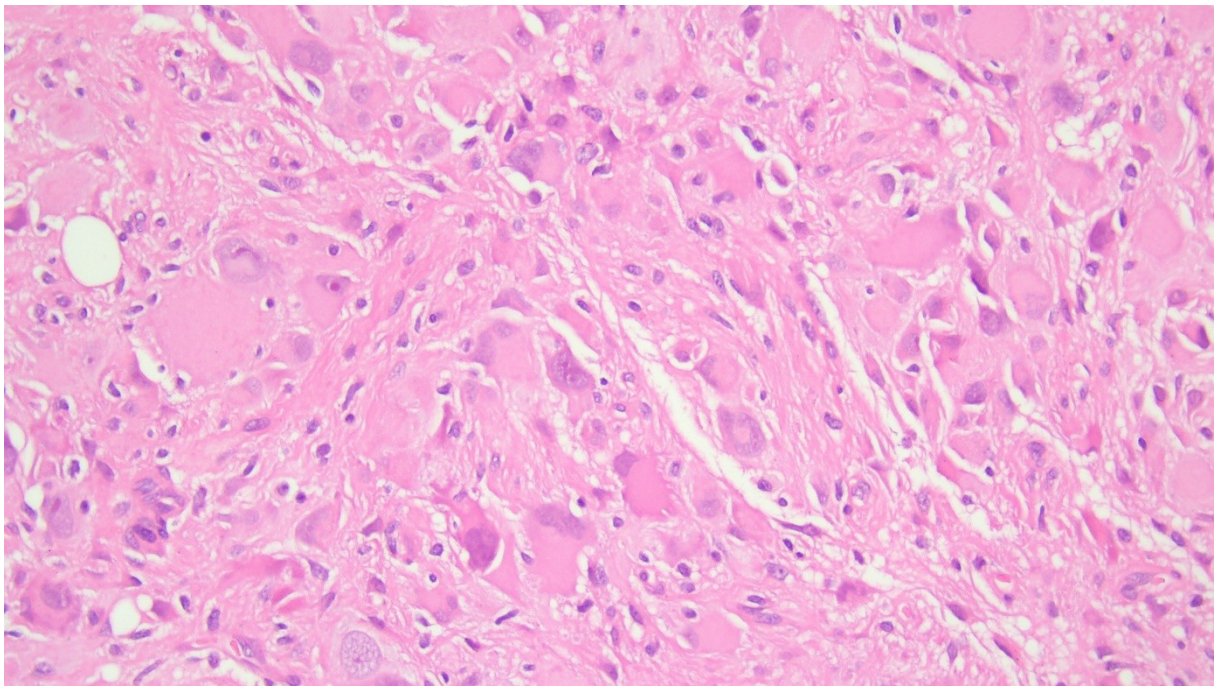

Figure 1. Photomicrograph (haematoxylin-eosin, 40x), showing a neoplasm consisting of epithelioid/ganglioid cells with a large cytoplasm and prominent nucleolus

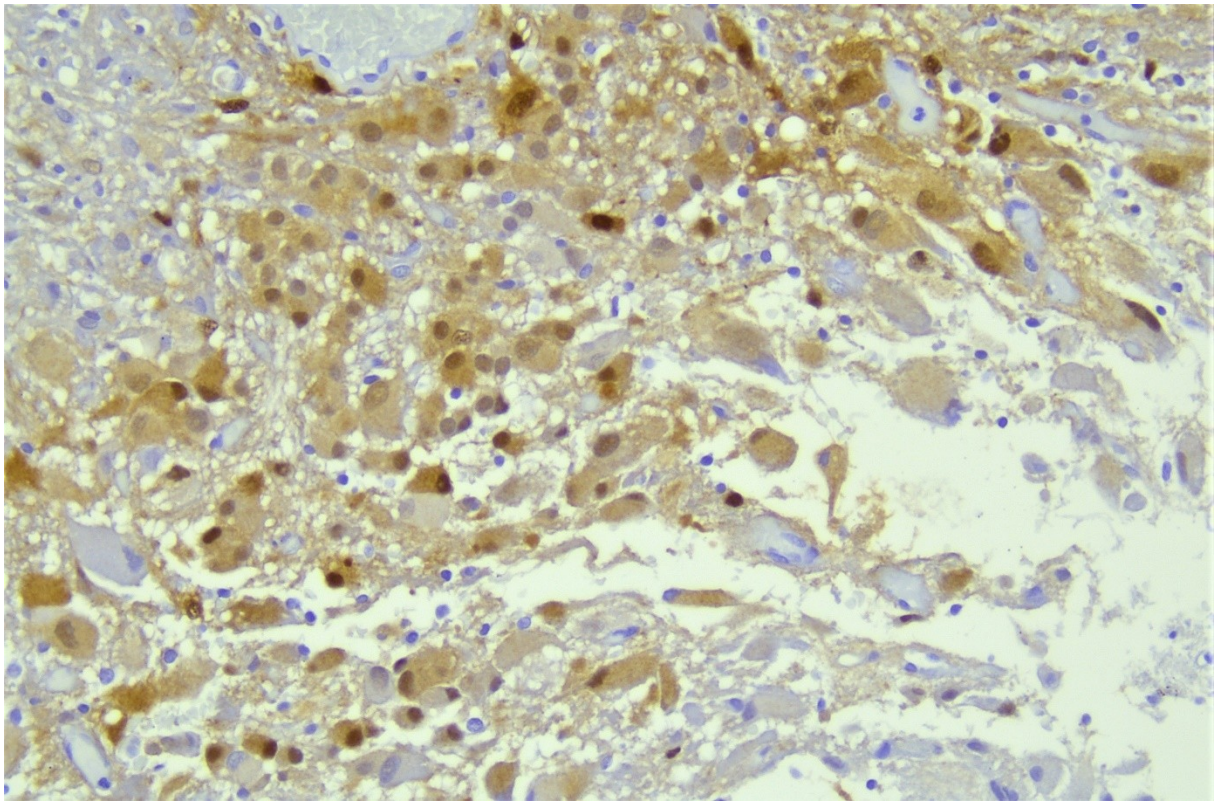

Figure 2. Immunohistochemistry showing positivity in the neoplastic cells for NTRK