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

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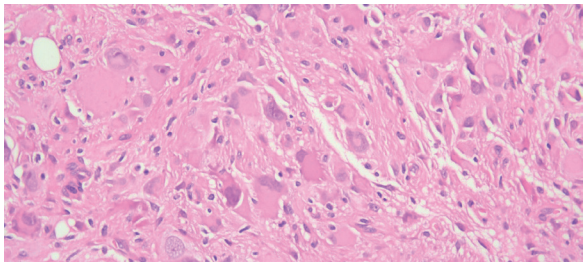

Figure 1. Photomicrograph (haematoxylin-eosin, 40x), showing a neoplasm consisting of epithelioid/ganglioid cells with a large cytoplasm and prominent nucleolus

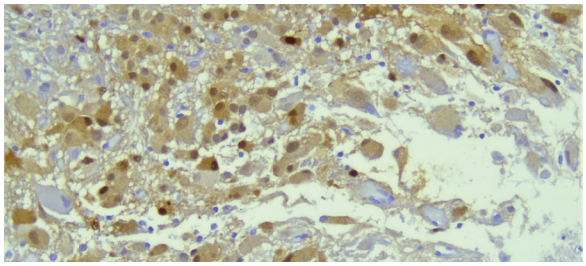

Figure 2. Immunohistochemistry showing positivity in the neoplastic cells for NTRK

An 11-year-old child presented with hydrocephalus-related symptoms. MR demonstrated, in the post T_1-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 the absence of mitosis and/or vascular proliferation and/or necrosis (fig. 1). Immunohistochemistry revealed positivity for GFAP and S-100. The diagnosis was SEGA – 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 WHO-CNS2016 and WHO-CNS2021 classifications 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 that are treatable with TRK-inhibitors, whereas our results suggest no correlation with immunocheckpoint expression.

References

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