Ependymoma RELA fusion positive

Ependymoma RELA fusion-positive is a accepted variant of ependymoma, only recognised in the World Health Organization Classification of Tumors of the Central Nervous System 2016.

Epidemiology

These tumors may account for 70% of supratentorial ependymomas in children and represent an aggressive entity distinct from other ependymomas.

Not found in the posterior fossa or spinal cord.

Two-thirds of supratentorial (ST) ependymomas harbor oncogenic fusions of RELA.

Outcome

The protein product is the principal effector of canonical Nuclear factor kappa signaling. RELA fusion proteins activate signaling for tumor proliferation and malignant progression, resulting in poorer prognoses in these patients compared to those in patients with other ST ependymomas.

In a study, Nakamura et al. encountered a case of C11orf-RelA fusion-positive ST anaplastic ependymoma that was diagnosed in first tumor resection surgery of multi-staged gross total resection with molecular evidence. In ependymomas, regardless of tumor location or pathological grade, subtotal resection is associated with higher rates of mortality compared with GTR.

In posterior fossa ependymoma group A (PF-EPN-A) tumors, telomerase activity varied and was significantly associated with dismal overall survival, whereas telomerase reactivation was present in all supratentorial RelA fusion-positive (ST-EPN-RELA) ependymomas.

Pathology

These tumours can be both grade II or III and demonstrate a variety of histological morphologies, although clear cells and prominent vascularity are common.

The presence of the RELA fusion gene can be assessed with FISH.

Immunophenotype

GFAP positive

EMA positive

L1CAM positive correlates closely with the presence of RELA fusion not exclusive to ependymomas.

Treatment

Actinomycin D could constitute a promising therapeutic option for Ependymoma RELA fusion positive, whose tumours frequently exhibit p53 inactivation.

Intraoperative MR perfusion imaging may be a useful method for delineating tumor aggressiveness and borders, which can be prognostic.
Case reports

Gamboa et al. from the Clinical Neurosciences Center, University of Utah, Salt Lake City, present the case of a patient with RELA-fusion positive ependymoma of the frontal lobe in whom they used preoperative and intraoperative MR perfusion imaging. In this first demonstrated intraoperative evaluation of MR perfusion in ependymoma, increased peripheral perfusion of the lesion in a ring-like manner with a discrete cutoff around the surgical margin correlated with intraoperative findings of a clear border between the tumor and brain, as well as pathological findings of increased MIB index and hypercellularity-specifically within solid tumor components. An abnormal perfusion pattern also suggested an aggressive lesion, which was later confirmed on pathological analysis. In addition, intraoperative MR perfusion improved detection of tumor tissue in combination with traditional T1-weighted contrast-enhanced methods, which increased extent of resection.

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2017

A case of aggressive anaplastic ependymoma arising in the right frontoparietal lobe, which had genetically 1q25 gain, CDKN2A homozygous deletion, and L1CAM overexpression. The patient was a 10-year-old boy who underwent four times of tumor removal and seven times of gamma knife surgery. Metastatic loci were scalp and temporalsis muscle overlying primary operation site, lung, liver, buttock, bone, and mediastinal lymph nodes. He had the malignancy for 10 years and died. This tumor is a representative case of Ependymoma RELA fusion positive, showing aggressive behavior.