Embryonal tumor with multilayered rosettes: diagnostic tools update and review of the literature.


Abstract

Embryonal tumor with multilayered rosettes (ETMR), including embryonal tumor with abundant neuropil and true rosettes (ETANTR), and ependymoblastoma (EBL) constitute a distinct entity of the primitive neuroectodermal tumor (PNET) family. The presence of a focal amplification at chromosome region 19q13.42 associated with an up-regulation of the oncogenic miRNA cluster C19MC suggests that they may represent a histological spectrum of a single biological entity. Their histopathological spectrum is wide, including medulloepithelioma, their location may be supra- or infra-tentorial, their prognosis is poor. Recent data on molecular subgroups of PNETs have led to new insights on diagnosis and treatment of these tumors. Subsequently, LIN28A immunoreexpression was identified as a highly specific marker for ETMR. In this study, we report 4 cases diagnosed initially as ETANTR with CGH-array data, including 19q13.42 gain with absence of other amplicons, particularly of the MYC gene family, and inconstant gain of whole chromosome 2. Immunohistochemical positive expression of LIN28A and absence of Olig2 expression were observed. We summarize the literature on ETMR, pointing out on the nosological evolution of this entity and the findings on genetic hallmarks of this particular tumor. Our results emphasize the usefulness of immunohistochemistry as a highly sensitive and fast diagnostic tool for ETMR and for genetic data, especially for 19q13.42 locus. Biological features may offer new therapeutic options for these embryonal tumors that do not usually respond to conventional treatments of PNETs.

PMID: 23863344 [PubMed - indexed for MEDLINE]

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