

PROVA ORALE N. I

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ATRX loss in adult gliomas lacking H3 alterations or IDH mutations, an exceptional situation for exceptional diagnoses: the experience of Sainte-Anne hospital

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Abstract

ATRX immunostaining constitutes a routinely used biomarker for the practice of neuropathology. The loss of ATRX expression correlating with *ATRX* gene alterations is implicated in a wide variety of pediatric and adult gliomas, and has been indexed as a desirable or essential diagnostic criterion for four tumor types featured in the latest world health organization classification of central nervous system Tumors. In adult-type diffuse glioma, the loss of ATRX expression is a hallmark of astrocytoma, IDH-mutant. Recently, novel tumor types and alterations have been referenced in the literature. These include the high-grade astrocytoma with piloid features (HGAP), for which no consistent clinicopathological features have been defined, and the presence of other alterations in the Krebs cycle genes (variants of the *Fumarate hydratase* -FH- gene) found in gliomas resembling astrocytomas, IDH-mutant. Because of this rapidly evolving classification and histomolecular landscape, we retrospectively analyzed adult gliomas diagnosed over a four consecutive year period to identify supratentorial gliomas, lacking H3 alterations or IDH mutations and harboring a loss of ATRX expression, in order to update their diagnoses in terms of histopathology, genetics and epigenetics. Four specimens (from 620 adult gliomas, 0.7%) were reclassified at the end of the molecular workup, as: 1/ one HGAP, 2/ one malignant transformation with a primitive neuronal component of an astrocytoma, IDH-mutant which lost the *IDH2* mutation at recurrence, 3/ a glioma, FH-mutant for which the histopathological and epigenetic features were similar to an astrocytoma, IDH-mutant, and 4/ a glioblastoma, IDH-wildtype. To conclude, these exceptional cases extend the spectrum of ATRX loss in gliomas, beyond the astrocytoma, IDH-mutant and the diffuse hemispheric glioma, H3 G34-mutant.

Keywords ATRX, Diffuse glioma, FH, HGAP

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