Fondazione I.R.C.C.S.
Istituto Neurologico Carlo Besta

Sistema Socio Sanitario
Regione Lombardia

PROVA ORALE N. 1

Fattori prognostici nei meningiomi.

MARIELLA CALLEA

MariellaCallea

6/08/2025

PROVA NON ESTRATTA

Fondazione I.R.C.C.S.
Istituto Neurologico Carlo Besta

Sistema Socio Sanitario
Regione
Lombardia

PROVA ORALE N. 2

Alterazioni molecolari possibili target terapeutici nei gliomi.

MARCELLA CALEA

Marcella Calea

06/08/2025

PROVA NON ESTRATTA

Fondazione I.R.C.C.S.
Istituto Neurologico Carlo Besta

Sistema Socio Sanitario
Regione Lombardia

PROVA ORALE N. 3

Principi classificativi dei tumori neuroendocrini dell'ipofisi.

MARCELLA CALLEA

Marcella Callea

06/08/2025

PROVA ESTRATTA

Fondazione I.R.C.C.S.
Istituto Neurologico Carlo Besta

Sistema Socio Sanitario
Regione Lombardia

QUESITO DI INFORMATICA

É possibile proteggere un documento pdf con password?

Tauziède-Espariat *et al.*
Acta Neuropathologica Communications (2025) 13:131
https://doi.org/10.1186/s40478-025-02044-6

Acta Neuropathologica Communications

RESEARCH Open Access

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

Arnault Tauziède-Espariat[1,2,3*], Alexandre Roux[2,3,4], Joseph Benzakoun[2,3,5], Paul Kauv[6], Sanaa Tazi[7], Alice Métais[1,2,3], Abigail K. Suwala[8,9,10], ·Felix Hinz[8,9], ·Lauren Hasty[1], Mathilde Filser[11], Julien Masliah-Planchon[11], Raphaël Saffroy[12], Margot Bucau[13], Johan Pallud[2,3,4] and Pascale Varlet[1,2,3]

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

*Correspondence:
Arnault Tauziède-Espariat
a.tauziede-espariat@ghu-paris.fr
Full list of author information is available at the end of the article