Pathol Res Pract. 2023 Dec:252:154942. doi: 10.1016/j.prp.2023.154942. Epub 2023 Nov 14.

Pilocytic astrocytoma in adults: Histopathological, immunohistochemical and molecular study with clinical association

Débora Salles [1], Samara F Santino [1], Paula Diana [1], Andréa C M Malinverni [2], João N Stávale [3]

Affiliations

PMID: 37984046 DOI: 10.1016/j.prp.2023.154942

Abstract

Pilocytic astrocytoma is the most common primary CNS neoplasm in children and adolescents, rare after the first two decades of life. While some authors report a favorable prognosis in the adult age group with the tumor, others have associated it with higher mortality. The molecular alteration most observed in cases of pilocytic astrocytoma in the pediatric group is the BRAF-KIAA1549 gene fusion, and there are still few studies confirming the presence of this fusion in the adult population. This work investigated genetic alterations involving the 7q34 region in BRAF gene in 21 adult individuals with pilocytic astrocytoma, by FISH. In addition, was identified the immunohistochemical expression of BRAFV600E, correlating these findings with histopathological and clinical ones. BRAF-KIAA1549 fusion appeared in only one case, while in two other cases were found deletions related to the FAM131B-BRAF fusion, suggesting that maybe the latter is more frequently in this population. Through the evaluation of immunoreactivity, 71% of the cases were considered positive and 29% negative. Cases considered positive for BRAFV600E immunoreactivity can potentially be treated through drug therapy with BRAF inhibitors; however, it is always recommended to carry out a molecular study for diagnostic confirmation. This is the first Brazilian study that aimed to investigate possible genetic alterations in the BRAF gene in pilocytic astrocytomas, specifically in adults. Only 1 patient died, but due to operative complications and not the disease itself, suggesting a good evolution of these individuals.

Keywords: Gene fusion; Immunohistochemistry; Molecular biology; Pathology; Pilocytic astrocytoma.