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Research paper 23

Testing for IDH mutation in astrocytoma by immunohistochemistry: an analysis of potential misdiagnosis in the Sri Lankan setting

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Introduction and objectives: The new World Health Organization Classification bases the diagnosis of astrocytoma on the presence of IDH mutation. Immunohistochemistry (IHC) for IDH mutation was introduced for the first time in our setup. This audit analyses the results of IDH testing by IHC and the potential for misdiagnosis.

Methodology: Twenty-two glial tumours were referred for analysis of IDH mutational status by IDH1 R132H IHC from January 2022 to July 2023. The histology was evaluated by two pathologists to confirm the diagnosis, grade and IDH status.

Results: Twenty tumours were high-grade (90.91%;20/22) and two were low-grade (9.09%;2/22). Among the high-grade tumours, IDH mutation was present in seven tumours including one with areas of oligodendroglial morphology. These were diagnosed as CNS WHO grade 4 astrocytoma with recommendations for IDH sequencing and 1p/19q codeletion in the tumour with oligodendroglial morphology. One of the tumours with low-grade histology showed IDH mutation and was reported as astrocytoma CNS WHO grade 2 with recommendations for IDH sequencing. The remaining thirteen high-grade tumours were IDH wild type and diagnosed as glioblastoma, WHO grade 4. One case of IDH wild type showed low-grade morphology.

Discussion: The lack of resources for molecular testing has led to several limitations in the diagnosis. IDH mutation should be tested in all glial tumours irrespective of grade, however, it is requested mainly in high-grade tumours. There are no facilities for testing of IDH wild-type low-grade tumours on IHC with IDH sequencing, to exclude false negatives. Facilities for testing for TERT promoter mutation, EGFR amplification, and combined chromosome 7 gain and chromosome 10 loss to exclude glioblastoma CNS WHO grade 4, in low-grade IDH wild-type tumours lacking microvascular proliferation or necrosis are also lacking. These limitations may result in the misdiagnosis of low-grade glial tumours.

Keywords: glioblastoma, IDH sequencing, oligodendroglia

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