

## HLAVNÍ TÉMA

PRAKTICKÁ DIAGNOSTIKA NÁDOROV CNS PODĚA WHO KLASIFIKÁCIE 2021: VŠEOBECNÉ ZMENY A DIAGNOSTIKA DIFÚZNYCH GLIÓMOV

9. Ellison DW, Aldape KD, Capper D, et al. cIMPACT-NOW update 7: advancing the molecular classification of ependymal tumours. *Brain Pathol.* 2020;30(5):863–866. doi: 10.1111/bpa.12866.
10. Ellison DW, Hawkins C, Jones DTW, et al. cIMPACT-NOW update 4: diffuse gliomas characterized by MYB, MYBL1, or FGFR1 alterations or BRAFV600E mutation. *Acta Neuropathol.* 2019;137(4):683–687. doi: 10.1007/s00401-019-01987-0.
11. Jaunmuktane Z, Capper D, Jones DTW, et al. Methylation array profiling of adult brain tumours: diagnostic outcomes in a large, single centre. *Acta Neuropathol Commun.* 2019;7(1):24. doi: 10.1186/s40478-019-0668-8.
12. Karimi S, Zuccato JA, Mamatjan Y, et al. The central nervous system tumor methylation classifier changes neuro-oncology practice for challenging brain tumor diagnoses and directly impacts patient care. *Clin Epigenetics.* 2019;11(1):185. doi: 10.1186/s13148-019-0766-2.
13. Keck MK, Sill M, Wittmann A, et al. Amplification of the PLAGL-family genes-PLAGL1 and PLAGL2-is a key feature of the novel tumor type CNS embryonal tumor with PLAGL amplification. *Acta Neuropathol.* 2023;145(1):49–69. doi: 10.1007/s00401-022-02516-2.
14. Louis DN, Giannini C, Capper D, et al. cIMPACT-NOW update 2: diagnostic clarifications for diffuse midline glioma, H3 K27M-mutant and diffuse astrocytoma/anaplastic astrocytoma, IDH-mutant. *Acta Neuropathol.* 2018;135(4):639–642. doi: 10.1007/s00401-018-1826-y.
15. Louis DN, Ohgaki H, Wiestler OD, Cavenee WK. World Health Organization Classification of Tumours of the Central Nervous System. 4th ed., updated ed. Lyon: International Agency for Research on Cancer. 2016. 408 s. ISBN 978-92-832-4492-9.
16. Louis DN, Perry A, Wesseling P, Brat DJ, et al. The 2021 WHO Classification of Tumors of the Central Nervous System: a summary. *Neuro Oncol.* 2021;23(8):1231–1251. doi: 10.1093/neuonc/noab106.
17. Louis DN, Wesseling P, Aldape K, et al. cIMPACT-NOW update 6: new entity and diagnostic principle recommendations of the cIMPACTUtrecht meeting on future CNS tumor classification and grading. *Brain Pathol.* 2020;30(4):844–856. doi: 10.1111/bpa.12832.
18. Louis DN, Wesseling P, Paulus W, et al. cIMPACT-NOW update 1: Not Otherwise Specified (NOS) and Not Elsewhere Classified (NEC). *Acta Neuropathol.* 2018;135(3):481–484. doi: 10.1007/s00401-018-1808-0.
19. Muench A, Teichmann D, Spille D, et al. A Novel Type of IDH-wildtype Glioma Characterized by Gliomatosis Cerebri-like Growth Pattern, TERT Promoter Mutation, and Distinct Epigenetic Profile. *Am J Surg Pathol.* 2023;47(12):1364–1375. doi: 10.1097/PAS.0000000000002118.
20. Pratt D, Abdullaev Z, Papanicolaou-Sengos A, Ketchum C, et al. High-grade glioma with pleomorphic and pseudopapillary features (HPAP): a proposed type of circumscribed glioma in adults harboring frequent TP53 mutations and recurrent monosomy 13. *Acta Neuropathol.* 2022;143(3):403–414. doi: 10.1007/s00401-022-02404-9.
21. Priesterbach-Ackley LP, Boldt HB, Petersen JK, et al. Brain tumour diagnostics using a DNA methylation-based classifier as a diagnostic support tool. *Neuropathol Appl Neurobiol.* 2020;46(5):478–492. doi: 10.1111/nan.12610.
22. Schepke E, Löfgren M, Pietsch T, et al. DNA methylation profiling improves routine diagnosis of paediatric central nervous system tumours: A prospective population-based study. *Neuropathol Appl Neurobiol.* 2022;48(6):e12838. doi: 10.1111/nan.12838.
23. Sievers P, Sill M, Schimpf D, et al. A subset of pediatric-type thalamic gliomas share a distinct DNA methylation profile, H3K27me3 loss and frequent alteration of EGFR. *Neuro Oncol.* 2021;23(1):34–43. doi: 10.1093/neuonc/noaa251.
24. Tauziède-Espariat A, Siegfried A, Nicaise Y, et al. PLAG1 fusions extend the spectrum of PLAG(L)-altered CNS tumours. *Acta Neuropathol.* 2023;146(6):841–844. doi: 10.1007/s00401-023-02643-4.
25. Tauziède-Espariat A, Volodia-Dangouloff-Ros, Figarella-Branger D, et al. Clinicopathological and molecular characterization of three cases classified by DNA-methylation profiling as „Glioneuronal Tumors, NOS, Subtype A“. *Acta Neuropathol.* 2022;144(6):1179–1183.
26. WHO Classification of Tumours Editorial Board. World Health Organization Classification of Tumours of the Central Nervous System. 5th ed. Lyon: 1. International Agency for Research on Cancer. 2021. 568 s. ISBN 9789283245087.

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### ODBORNÝ GARANT:

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