

<https://doi.org/10.7124/bc.000B76>

**S.S. Kalman¹, L.M. Kovalevska¹, A.V. Suchnova¹,
M.V. Tomiak², R.V. Gulkowskyi², O.Yu. Malysheva³,
T.A. Malysheva³, E.V. Kashuba¹**

¹ R.E. Kavetsky Institute of Experimental Pathology, Oncology
and Radiobiology, NAS of Ukraine
45, Vasylykivska Str., Kyiv, Ukraine, 03022

² Institute of Molecular Biology and Genetics, NAS of Ukraine
150, Akademika Zabolotnoho Str., Kyiv, Ukraine, 03143

³ SI "Romodanov Neurosurgery Institute National Academy
of Medical Sciences of Ukraine"
32, Platon Maiboroda Str., Kyiv, Ukraine, 04050
kalmansergej12@gmail.com

MOLECULAR PROFILING OF BRAIN TUMORS USING NEXT-GENERATION SEQUENCING: IDENTIFICATION OF CLINICALLY RELEVANT MUTATIONS

Background/Aim. A five-year survival rate rarely exceeds 35% for brain tumors. Hence, there is an urgent need for improved molecular diagnostics. Here we performed molecular profiling of brain tumor samples by identifying clinically relevant point mutations. **Methods.** To do so, we used the Ion AmpliSeq Cancer Hotspot Panel v2 (~2800 hotspot regions across 50 cancer-related genes) on an Ion GeneStudio S5 Plus platform and an Ion GeneStudio S5 Plus Sequencer machine (Thermo Fisher Scientific) to analyse genomic DNA isolated from 32 brain tumor tissue samples. **Results.** In glioblastoma G4, the characteristic IDH1 Arg132His substitution was detected alongside with inactivating TP53 mutations (Met246Ile, Tyr125Met, Cys135Trp). In diffuse astrocytoma G2 the BRAF Val600Glu mutation – a known therapeutic target – was found, together with the FGFR1 Lys145Gln and GNAQ Gln209Lys mutations. Atypical meningioma G2 harbored the AKT1 Glu17Lys mutation, causing constitutive activation of the PI3K/mTOR pathway. In medulloblastoma G4, four mutations were identified: APC Ala158Thr and CTNNB1 Gly34Arg (disrupting the WNT pathway), FGFR3 Val672Ile, and PIK3CA Gly1049Arg (constitutively activating the PI3K pathway). In embryonal tumors of the CNS G4 the BRAF Val600Glu, GNAS Asp229Asn, RB1 Met708Thr (partially inactivating the pocket domain critical for E2F interaction), and SMO Phe403Leu (conferring potential resistance to vismodegib/sonidegib) were detected. **Conclusions.** Ion Torrent-based NGS with a cancer hotspot panel enabled efficient and cost-effective identification of clinically significant mutations across diverse brain tumor types in a single sequencing run. Several detected mutations, including BRAF Val600Glu, AKT1 Glu17Lys, and SMO Phe403Leu, have direct therapeutic implications, confirming the value of molecular profiling for treatment optimization. These findings support routine integration of a NGS-based hotspot analysis into the diagnostic workflow for brain tumor patients. **Grants/Fundings.** This work was supported by a research program of National Academy of Sciences of Ukraine "Study on molecular mechanisms of growth inhibition of embryonic tumors upon inactivation of the MRPS18–2 protein" (0123U100099).

Keywords: brain tumors, next-generation sequencing, Ion Torrent, molecular profiling, cancer hotspot mutations, glioma.

Received: 06.07.2026; Accepted: 08.09.2026; Published: 29.09.2026

Citation: Kalman S.S., Kovalevska L.M., Suchnova A.V., Tomiak M.V., Gulkowskyi R.V., Malysheva O.Yu., Malysheva T.A., Kashuba E.V. (2026) Molecular profiling of brain tumors using next-generation sequencing: identification of clinically relevant mutations. *Biopolymers & Cell*, Special issue(42), 59. <https://doi.org/10.7124/bc.000B76>

© Publisher PH "Akademperiodyka" of the NAS of Ukraine, 2026. This is an Open Access article distributed under the terms of the Creative Commons Attribution License (<http://creativecommons.org/licenses/by/4.0/>), which permits unrestricted reuse, distribution, and reproduction in any medium, provided the original work is properly cited