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Genomic landscape of glioblastoma without IDH somatic mutation in 42 cases: a comprehensive analysis using RNA sequencing data

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Abstract

Purpose: Glioblastoma is a malignant brain tumor with a poor prognosis. Genetic mutations associated with this disease are complex are not fully understood and require further elucidation for the development of new treatments. The purpose of this study was to comprehensively analyze genetic mutations in glioblastomas and evaluate the usefulness of RNA sequencing.

Patients and methods: We analyzed 42 glioblastoma specimens that were resected in routine clinical practice and found wild-type variants of the IDH1 and IDH2 genes. RNA was extracted from frozen specimens and sequenced, and genetic analyses were performed using the CLC Genomics Workbench.

Results: The most common genetic alterations in the 42 glioblastoma specimens were TP53 mutation (28.6%), EGFR splicing variant (16.7%), EGFR mutation (9.5%), and FGFR3 fusion (9.5%). Novel genetic mutations were detected in 8 patients (19%). In 12 cases (28.6%), driver gene mutations were not detected, suggesting an association with PPP1R14A overexpression. Our findings suggest the transcription factors SOX10 and NKX6-2 are potential markers in glioblastoma.

Conclusion: RNA sequencing is a promising approach for genotyping glioblastomas because it provides comprehensive information on gene expression and is relatively cost-effective.

Keywords: Fusion gene; Gene expression; Glioblastoma; RNA sequence.

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