Copy number profiling of Brazilian astrocytomas

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Copy number alterations (CNA) have been currently used as important biomarkers in gliomas' routine setting. Therefore, the aim of this study was to characterize the genomic profile of astrocytomas of Brazilian origin in order to describe targets to be object of future investigation. For this, we performed CNA profiling of 65 astrocytomas of distinct malignant grades, using array-CGH, microsatellite instability analysis (MSI), and investigated their correlation with *TERT* and *IDH1* mutational status and clinic-pathological features. Finally, *in silico* analysis using Oncomine database was performed to validate our findings and extend the findings to gene expression level. We found that, in glioblastomas (GBM) the most common alterations were amplifications of *PDGFRA*, *KIT, KDR*, *EGFR* and *MET,* and deletions of *CDKN2A* and *PTEN*. Log-rank analysis correlated *EGFR* amplification and/or chr7 gain with better survival of the patients. MSI was observed in 11% of GBMs. A total of 69% of GBMs presented *TERT* mutation, whereas *IDH1* mutation was most frequent in diffuse (85.7%) and anaplastic (100%) astrocytomas. The combined analysis of 1p19q deletion, *TERT* and *IDH1* mutational status separate tumor groups that showed distinct age of diagnosis and outcome. *In silico* validation pointed to less explored genes that may be worthy of future investigation, such as *CDK2*, *DMRTA1* and *MTAP*. Besides showing that the genomic profile in Brazilian population was similar to the described worldwide, we indicated potentially important genes, not extensively studied in gliomas, which could be further explored to assess their biological and clinical impact in astrocytomas.

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