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Molecular genetics assay development using next generation DNA sequencing for mutation detection in brain tumors

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Molecular pathology and molecular diagnostic areas are slowly progressing in the Kingdom of Saudi Arabia. Molecular diagnostic testing boosts therapeutic products use in a more precise way to treat cancer and other diseases also. Until recently, routine screening of tumors for mutations was confined to capillary sequencing or Real-Time PCR. However, in recent years molecular pathology laboratories are adopting Next Generation Sequencing (NGS) platforms for routine diagnostic screening of solid tumors. The detection of exact genetic anomalies can help in predicting prognosis and guide the selection of targeted therapies. Our aim is to develop the Next Generation DNA Sequencing in brain tumor samples as a strategy for molecular genetic test development in Saudi Arabia. DNA samples from glioblastoma and ependymoma cases were analyzed by Ion Proton sequencing using Ion AmpliSeq Cancer Hotspot panel v2 primers. In grade III ependymoma, we found 11 exonic (8 synonymous and 3 missense), 8 intronic and one 3'-UTR variants. In this case missense mutations detected were in KDR c.1416A>T; in PIK3CA c.1173A>G; and in TP53 c.215C>G; respectively. Coverage data analysis showed that 99.52% and 93.24% amplicons had at least 100 and 500 reads respectively. Target base coverage at 100x was 99.61% and 93.16% at 500x coverage for this. In glioblastoma case, we found missense mutations such as c.373G>A in KRas, c.899C>T in TP53, c.1715C>T in JAK3, c.352C>T in IDH1, c.2758C>G in ERBB4, c.1573G>A in PIK3CA, c.2320G>A in FGFR3, c.1787C>T in EGFR genes. Also, nonsense mutations in BRAF c.1349G>A, and PTEN c.19G>T, were detected in glioblastoma patient. The development and application of a genetic test for glioma markers at molecular level using NGS technologies will have significant economic benefits for the Kingdom of Saudi Arabia.

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