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A novel infram deletion in *MSH6* gene in glioma: Conversation on *MSH6* mutations in brain tumors

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Background & Aim: Histological and molecular information and biopsy help in the diagnosis of the type and grade of tumors and increase the value of estimation of the biological behavior of tumors. In this study, we aim on a consanguineous Iranian family with high prevalence of brain tumors in their pedigree and reviewed the literature on *MSH6* mutations in brain tumors and the treatment responses focused on gliomas.

Method: We chose a family with a high prevalence of brain tumor in their pedigree. We studied the proband's neuroimaging and brain proton Magnetic Resonance Spectroscopy (MRS), Magnetic Resonance Imaging (MRI), biopsy result and whole genome sequencing.

Result: The neuroimaging and brain proton MRS reported a lesion in the right frontoparietal. The MRI revealed a large enhancible heterogenous mass in the right temporo-fronto-parieto-occipital lobes with involvement of corpus callosum which was suggestive of glioma. The patient revealed a homozygous pattern for a novel 9 base-pare deletion at the 912-914 codon on exon 4 of the *MSH6* gene.

Discussion: We discuss several studies on *MSH6* mutations in brain tumors and we discuss treatment responses in *MSH6* mutations and the studies conducted to sensitize chemotherapy and radiotherapy resistance brain tumors to face this subject efficiently.

Conclusion: Patients should be evaluated for MMR mutation before chemo and radiotherapy, and it is valuable to follow-up these mutations during the treatment too. In Temozolomide (TMZ)-resitance cases, it is suggested to use complementary strategies such as using HDAC is and a combination of a STAT3 Inhibitor and an mTOR inhibitor, BER inhibition mechanism and PARP-1 inhibitor. The highlights of this study are central nervous system tumors are rare and they are responsible for approximately 20-30% of cancer morbidity in children and youngsters, Mismatch Repair (MMR) mutations and mutations in the DNA-repair system are probably related to tumor progression and chemo and radiotherapy resistance, germline mutations of *MSH6* increase the susceptibility to gliomas and somatic mutations cause temozolomide (TMZ)-resistance manner, inhibition of NAD+ biosynthesis increases the expression of MGMT and this can support the deficiency of MMR proteins. Making a connection between the genetic profiles and the evolved pathways and therapeutic approaches enhance our vision in estimating the results beyond treatment.

Biography

Zeinab Deris Zayeri has completed her Msc in Human Genetic from Genetic department, Ahvaz Jundishapur University of Medical Sciences, Ahvaz, Iran. She is the Head of Clinical Labratory in Ahvaz. She has published more than 20 papers in reputed journals and has been serving as an Editorial Board Member of *Cancer Research Journal*.

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