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The Effect of Catalase Enzyme Gene Polymorphism A-21T (rs7943316) on Epilepsy and its Drug Resistance after Hypoxic Ischemic Brain Injury

Background: Epilepsy is one of the most common serious neurological disorders, affecting more than 4% of all children. One of the most common conditions leading to epilepsy is Hypoxic ischemic encephalopathy (HIE) which is a condition that occurs when the entire brain is deprived of an adequate oxygen supply. Catalase (CAT) is a major cytoplasmic antioxidant enzyme. Considering that the A-21T and C-262T polymorphisms in the promoter region of CAT are associated with the activity of promoter of the CAT; subsequently it may alter the risk of oxidative stress related disorders. Therefore, polymorphism of the CAT gene can be a candidate marker of the risk of epilepsy.

Objectives: To assess if antioxidant CAT gene polymorphism A-21 T (rs7943316) contributes to susceptibility to epilepsy, susceptibility to epilepsy after neonatal HIE, susceptibility to epilepsy due to other causes than neonatal HIE, resistance to antiepileptic medications in epileptic patients after HIE, resistance to antiepileptic medications in patients due to other causes than HIE.

Methods: This cross sectional case-control descriptive analytical study included 105 subjects; 70 patients suffering from epilepsy (divided into two groups according to the etiology of epilepsy) compared to 35 age and sex matched healthy control subjects. The patients were recruited from neuro-pediatrics clinic in Fayoum University Teaching Hospital during a period extending from September, 2017 till February, 2018. All samples were subjected to genomic DNA analysis catalase enzyme polymorphism A-21T (rs7943316) using real time polymerase chain reaction based method.

Results: Our study showed that there was a statistically significant difference (p-value <0.05) between patients with epilepsy due to HIE and controls as regard genotyping where AA (wild genotype) was higher among controls, while AT (heterozygous mutant genotype) was higher among cases. Also AT (heterozygous mutant genotype) and T allele were statistically significantly higher among epilepsy with HIE cases when compared to epilepsy without HIE (p-value <0.001 and < 0.01 respectively). But there was no statistically significant difference in CAT rs7943316 genotype and allele frequency when epilepsy subjects were stratified by drug resistance, electroencephalography or by gender.

Conclusion: Our study revealed that there was a significant link between CAT A-21T (rs7943316) single nucleotide polymorphism (SNP) and

susceptibility to epilepsy after neonatal HIE. CAT polymorphism does not influence the overall risk of drug resistance among subjects with epilepsy after neonatal HIE or due to other causes than HIE.

Keywords: Epilepsy, Hypoxic ischemic encephalopathy, catalase, single nucleotide polymorphism