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Role of aminoacyl-tRNA synthetases (AARS) gene in epileptic encephalopathy, early infantile, 29 in Pakistani population

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Epilepsy is one of the most common neurological disorders and is more common in developing countries as compared to developed one. Epilepsy has a various type of phenotype depends upon the pathways involved in the signaling process. Epilepsy is a group of neurological disorders characterized by recurrent epileptic seizures. Genetics is believed to be involved in the majority of cases. During the last decade, many genes and mutations associated with epilepsies have been identified. To find out the genuine cause of the epilepsy in Pakistani idiopathic epilepsy patients was the major factor behind this project. Five patients suffering from early infantile were selected on the basis of the clinical findings. Although it is understood that SCN1A gene is the most causative factor for genetic epilepsies and majority of the cases of early infantile are found to be responsible by SCN1A gene. In current study Central Punjab from Pakistan was selected for the identification of epilepsy patients to conduct molecular study for now and future. Epilepsy questionnaire (from NINDS) was used which is the short form of the questionnaire. Whole Exome sequencing was the key technique to find out the molecular alterations. Aminoacyl-tRNA synthetases are indispensable enzymes in protein production because they allege tRNAs with their cognate amino acids. In this study we found that epileptic encephalopathy, early infantile, 29 is caused by the mutation in AARS gene.

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