Abstract ID: - 125

Abstract Topic: - Molecular effects of genetic variation

Abstract Title: - p.V1393M in CACNA1A: A Genetic Link to Epilepsy?

Presenting author name: - Shruti Anand

Presenting author institute: - MGM School of Biomedical Sciences (SBS)

Co-authors name: - Dr. Anjali Sabnis, Dr. Yogesh Patil, Dr. Virti Shah, , , , , ,

Co-authors institute: - MGM Medical College, MGM School of Biomedical Sciences (SBS), MGM Hospital, , , , , ,

Aims: - This research aimed to study the frequency of p.V1393M variation on exon 26 of the CACNA1A gene to understand its phenotypic effects with respect to epilepsy using PCR-SSCP.

Methods: - For the purpose of this study, subjects with epilepsy associated with ataxia, hemiplegic migraine, status epilepticus, or development delay will be screened and selected. Post-selection 2 mL blood samples will be collected for DNA extraction and then the extracted samples will be utilized for variant screening of the p.V1393M present on the CACNA1A gene. Post analysis the statistical significance of the prevalence will be evaluated for testing of the proposed hypothesis.

Results: - Proposed research attempts are being made to detect the prevalence of p.V1393M present on the CACNA1A gene and reveal the phenotypic symptoms in the epileptic population of the Konkan district of Maharashtra.

Conclusions: - According to the literature, this variation is prevalent in the Chinese population. An attempt is being made to replicate similar studies in the epileptic population of the Konkan District of Maharashtra with the hypothesis that a similar outcome will be observed.

Keywords: - Proposed research attempts are being made to detect the prevalence of p.V1393M present on the CACNA1A gene and reveal the phenotypic symptoms in the epileptic population of the Konkan district of Maharashtra.