

33<sup>rd</sup> Conference on  
Clinical Neuroscience and Neurogenetics  
March 25, 2022 | Webinar

### Genetics of photoparoxysmal response in idiopathic generalized epilepsy

Photoparoxysmal response (PPR) is an abnormal cortical response to photic stimulation manifesting as specific EEG changes. It is observed in 10% of idiopathic generalized epilepsy (IGE). The aim of this study is to understand the molecular genetics of Grade IV PPR, which is characterized by generalized spike and wave or polyspike and wave discharges. Fourteen children with IGE, exhibiting Grade IV PPR, were recruited for the study. Whole exome sequencing (WES) was done using SureSelectXT Human All Exon v5+UTRs kit on HiSeq 2500. Seventy six variants were observed in all the 14 samples. Among these, 65 variants were of missense, nonsense or frameshift types. Twelve of these variations were predicted to be deleterious. Eleven genes are novel candidate genes of epilepsy. The genes that had variations in all samples were enriched into the following biological processes (FDR <0.05), (i) development and functioning of the nervous system, (ii) axon guidance, (iii) NCAM1 interactions, (iv) LICAM interactions (v) laminin interactions (vi) NCAM signaling for neurite outgrowth. Dysfunction of these biological processes can lead to faulty neurodevelopment. Studies have shown that several developmental factors such as, altered neuronal signaling during embryonic life, defects in postnatal maturation of neuronal networks, and congenital brain malformations contribute to epileptogenesis. Hyperexcitability of neural network is a key neurophysiological mechanism in epilepsy. Migration defects of excitatory and inhibitory neurons and perturbations in the developmental refinement of neuronal circuitry during critical periods of neurodevelopment may trigger hyperexcitability and epilepsy later in life.

**Anitha Ayyappan Pillai**

*Institute for Communicative and  
Cognitive Neurosciences, India*

#### Biography

Dr. **Anitha Ayyappan Pillai** did her Ph.D. at Rajiv Gandhi Centre for Biotechnology, Trivandrum in the field of **Population Genetics**. She then worked as a Postdoctoral fellow and then as Assistant Professor at **Hamamatsu University School of Medicine**, Japan. At present, she works as an Associate Professor at Institute for Communicative and Cognitive Neurosciences (ICCONS), Shoranur. Her main research area is **Neurogenetics**. Dr. Anitha has received research grants from national and international funding **agencies** in India and Japan. She has authored >40 scientific papers in leading international journals and has co-authored book chapters.

**Received:** February 01, 2022; **Accepted:** February 02, 2022; **Published:** March 25, 2022