Novel candidate genes for autism spectrum disorders identified by whole exome sequencing of Indian autism twin, triplet and quadruplet families

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Autism spectrum disorder (ASD) is a childhood-onset complex neurodevelopmental disorder with a complex genetic architecture. To identify the potential candidate genes of ASD, we carried out a whole exome sequencing (WES) study of ASD twin, triplet and quadruplet families in the Indian population. Five monozygotic twin-, five dizygotic twin-, one monozygotic triplet-, and one multizygotic quadruplet- families participated in the study. The monozygotic twins, triplet and quadruplet were discordant for ASD, while the dizygotic twins were concordant for ASD. WES was done for all the members of each family. The de novo and inherited variants of probands were filtered from WES data. Among the proband-specific de novo and inherited variants, there were 23 deleterious variants. Some of the novel ASD candidate genes include, TRAM2, DGKD, OR5AC2, FLNB, TENM2 and ADAMTS18. These genes are known to play crucial roles in neurodevelopment, axon guidance and synaptic pasticity. They have been implicated in the pathogenesis of neurobehavioral disorders such as epilepsy and Ehlers–Danlos syndromes that share genetic etiologies and biological processes with ASD. Gene ontology enrichment analysis showed that the genes harboring proband-specific variants were enriched in biological processes involving cell adhesion, synaptic transmission and nervous system development. Embryonic neurogenesis is considered as a potentially important period in the pathogenesis of ASD. The onset of ASD is early in life during the period of rapid synaptogenesis. Aberrations in the aforementioned biological processes may lead to altered neurogenesis causing ASD. This is the first comprehensive genetic study of ASD in any Indian population.

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.

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