Population and family based association study on TPH1, TPH2 and ITGB3 genes indicate serotonergic system involvement in autism spectrum disorder

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Autism spectrum disorder (ASD) is one of the most common genetically predominant neurodevelopmental disorders and the prevalence rate is more than one percent. The pathophysiology of the disorder is highly complex and several genes are believed to involve with each having minor contribution leading to the final ASD phenotype. In the last two decades, various risk variants of different genes have been identified but none of the individual variants account for more than 1% of ASD cases in average. Recently, identification of genes that contribute to specific autism endophenotype is highly appreciated. So far two most common finding in ASD research are transient increase of head circumference and platelet hyperserotonemia. And platelet hyperserotonemia has been considered as autism endophenotype. Moreover, abnormal serotonin synthesis capacity has been implicated in the brain of ASD individuals, thereby the involvement of serotonergic system abnormality both in the brain as well as periphery in the pathophysiology of ASD. Investigation of few selected single nucleotide polymorphisms from three potential serotonergic candidate genes, TPH1, TPH2 and ITGB3 via population and family based approaches, revealed that these genes are moderately risk to the disorder. Gene-gene interaction analysis further suggests their interactive role towards the cause of ASD. However, as we do not have the functional experimental data, the present finding provides only preliminary evidence. However, these data highlight the need of extensive research both in genetic and functional level for clear understating the role of these genes in the etiology of ASD.

Biography
Asem Surindro Singh is presently working as Postdoctoral Fellow in Dr. Axel Brockmann’s Lab at NCBS. He completed his PhD research in May 2013 in the University of Calcutta under the supervision of Dr. Usha Rajamma. His main research interest is to understand the mechanism of how genes regulate behaviors through certain neural circuitry systems.

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