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Tryptophan hydroxylase gene involvement in the serotonergic abnormality of autism spectrum disorders: A genetic and genotype–phenotype correlation study

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One of the most consistent finding in ASD research is the presence of high serotonin level in the blood platelets of 30% ASD subpopulations. Abnormal serotonin synthesis capacity has also been implicated in the brain of ASD individuals. Even though serotonergic pathway in brain and the periphery is differentially regulated, during early stage of life when the blood brain barrier is not fully formed, the only rate limiting enzyme available for serotonin biosynthesis is the peripherally expressed *TPH1* which is the isoform of brain specific TPH2. On the light of these evidences the present study is a thorough investigation of *TPH1* gene to find the risk locus for serotonergic abnormality in ASD. From the family based approaches with individual markers, we found that A allele of rs10488682 is significantly overtransmitted from parent to female probands. Two-locus haplotype analyses showed significant overtransmission with various combinations of different SNPs that include rs623580, rs1800532, rs684302, rs1799913, rs211106 and rs10488682. Similar observation was also found with three locus haplotype models. Functional prediction analysis illustrated polymorphisms of these markers have very low to moderate effect on the gene regulation. Investigation with platelet serotonin assay showed that 33% of ASD individuals are hyperserotoninemic in our study population in India and is very close to the earlier reports of 30% (approx). Furthermore, quantitative trait association analyses with specific alleles, genotypes and haplotypes also demonstrated involvement of rs684302, rs211106, rs1800532 and rs1799913 of *TPH1* in the increased platelet serotonin level of ASD. Thus our various approaches with genetic association studies and genotype-phenotype correlation analyses suggest involvement of *TPH1* in the pathophysiology of ASD through the regulation of serotonin level. Since we do not have the functional experimental data, our findings provide the preliminary evidences. However, these data highlight the need of extensive research both in genetic and functional level for clear understating of *TPH1* involvement 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. His main research interest is to understand the mechanism of how genes regulate behaviors through certain neural circuitry systems.

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