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ASSOCIATION OF OXYTOCIN RECEPTOR (OXTR) GENE POLYMORPHISMS WITH AUTISM SPECTRUM DISORDER (ASD): A CASE-CONTROL STUDY WITH CLINICAL PROFILING

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Autism spectrum disorder (ASD) is a group of sex biased neurodevelopmental disorders characterized by core deficits in social behavior, communication and behavioral flexibility. Several lines of evidence indicate that oxytocin signaling through its receptor (OXTR), is essential in a wide range of social behaviors. Variable clinical profile of ASD with unique presentation of symptoms was found in our study. 92.23% of the ASD patients (n=103), than the control groups (n=30) were found to have significant ($p<0.0001$) social interaction difficulties. Along with that common developmental concerns significantly associated with language impairments ($p<0.001$), behavioral abnormalities like hypo or hyperactivity ($p<0.003$), repetitive behavior, resistance to change ($p<0.001$), self-hurting activities ($p<0.0001$). Co-morbidities as depression ($p<0.01$), dyslexia ($p<0.0001$), intellectual disability ($p<0.001$), sleep disturbance ($p<0.001$) are found to be significantly associated with ASD, which is co-related with anxiety and behavioral problems. Early recognition of symptoms and the risk factors would help in appropriate therapeutic intervention resulting in favorable outcome. In attempts to determine whether genetic variations in the oxytocin signaling system contribute to ASD susceptibility, we have investigated the role of OXTR variants in ASD development in our Bengali of Bangladesh (BEB) population of Chittagong region by analyzing four OXTR variants (rs53576, rs2254298, rs2228485, rs237911) through PCR-RFLP method based on case-control study (37 cases, 15 controls). A significant ($p<0.05$) frequency for OXTR rs53576 AA risk allele was found to be associated with ASD compare to controls which is consistent with the previous study in Chinese but Caucasian and Japanese population. No significant association has been found for OXTR variants (rs2254298, rs2228485, rs237911) in this study. These findings suggested to further investigate in a larger sample size on OXTR rs53576 polymorphism towards ASD in overall BEB population as well as ethnic group to open new avenue for clinical marker development for ASD diagnosis and treatment.

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