MOLECULAR SCREENING FOR 15Q11-Q13 DUPLICATIONS/DELETIONS IN INDIVIDUALS WITH AUTISM SPECTRUM DISORDER IN LAGOS METROPOLIS


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Introduction: Autism spectrum disorder (ASD) is a neurodevelopmental disorder, characterized by impairments in communication skills, social interaction, restricted or stereotyped patterns of behaviors. Different chromosomal aberrations have been linked to the aetiology of autism. Research studies have associated duplications and or deletions to chromosome 15q11-q13 in autism individuals. In most developed countries ethnically diverse and large sample sizes have been used in autism research, but very little representations have emerged from the African population. It is pertinent to present findings from an understudied population just to give a more global view on Autism spectrum disorders (ASD).

Objective: To screen for duplications and or deletions of 15q11-q13 in autism individuals from an understudied African population.

Materials & Methods: Blood samples were collected from consented participants at the Department of Paediatrics, College of Medicine, University of Lagos and Child and Adolescent Centre, Federal Neuro-Psychiatric Hospital, Lagos, Nigeria. DNA was isolated from the blood samples of probands and their available family members according to manufacturer’s instructions.

Five microsatellite markers (D15S156, D15S97, D15S975, D15S122, and D15S128) specific for chromosome 15q11-q13 region were used to screen for duplications and or deletions on the probands of autism children and their family members. Resulting PCR products were separated on 2% agarose gel electrophoresis.

Results: Maternally derived duplications and deletions were more prominent (>80%) in autism children than the paternally derived duplications and deletions for different markers used. Among the proband samples, duplication was found to be more for the GABRG3 gene that is amplified by the marker D15S975 with a percentage of 22.58%, while 51.61% deletions were observed within the ubiquitin protein ligase E3A (UBE3A) gene on the D15S122 and the OCA2 gene on the D15S156 marker regions.

Conclusions: Duplications and or deletions of 15q11-q13 that are maternally derived may play an important role in the genetic aetiology of autism.

Keywords: Autism Spectrum Disorders (ASDs), chromosomal aberration, Molecular screening.