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Genomic determinants of congenital mental retardation assorted through linkage analysis with STR markers in consanguineous families

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Abstract

Linkage analysis through microsatellite markers is a powerful tool to identify the disease causing variants in consanguineous families. About 1-3% of the human population suffers from learning and adaptive disabilities; termed as mental retardation (MR). One half of it is attributed to genetic factors and is therefore inheritable. Genetic basis of mental retardation is poorly understood and work emphases have been done X chromosome linked variants. Consanguineous populations are highly probable for genetic disorders and have an increased likelihood of rare, recessive disease causing variants. This in view, twenty families having multiple affected members segregating MR in autosomal recessive pattern of inheritance were enrolled. DNA was subjected to linkage analysis studies by genotyping with short tandem repeat (STR) markers for reported autosomal recessive nonsyndromic MR (arns-MR) loci. Nonsyndromic MR has been defined by the presence of intellectual disability as the sole clinical feature and OMIM uses the acronym MRT for arns-MR loci. Five families were found linked to reported MRT loci at chromosome 4 (two families), 19, 1 & 5 (one to each). The linkage intervals for these families have reduced the reported domains leading to a step forward for finding genetic variants. While unlinked families leave a gap to investigate the genomic determinants for this disorder. This information will be helpful and upshot for researchers to develop new diagnostic and prognostic tools for carrier status and prenatal diagnosis in affected population and sporadic cases too. The linkage data is also helpful in establishing genotype-phenotype correlation and an addition in data bank for this disorder.

Biography

Muhammad Yasir Zahoor, obtained his Ph.D. on molecular genetics of neuro-developmental disorders in 2011 and currently working as Assistant Professor of molecular biology in Pakistan and has research interest in genetic disorders.

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