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Usefulness of chromosomal microarrays in diagnosing genetic disease in UAE

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Introduction: Chromosomal microarray (CMA) is frequently performed as part of the diagnostic work-up for children with dysmorphic features, developmental delay, growth failure, and congenital anomalies. The traditional microarrays were designed for detection of gene copy number variations (CNVs) by comparative genomic hybridization (CGH), and were popularly called array CGH. The second type of arrays containing a high density of probes for single nucleotide polymorphisms (SNPs) reveals both CNVs and absence of homozygosity (AOH) areas in the genome. AOH areas can be explained by consanguinity or uniparental disomy (UPD). Previous studies has demonstrated 10-15% detection rate for pathogenic CNVs using CMA. The role of CMA in diagnosing genetic disorder in highly consanguineous community such as in UAE has not yet been described. Herein, we present the yield of CMA performed at the Tawam Hospital, Al-Ain, UAE during one year period.

Methods: The results of CMA performed in the Genetic Clinics in Tawam Hospital during the period from November 2014 to October 2015 were collected. The CMA tests, which used both SNP and array CGH methodologies, were performed in a certified clinical lab in USA. We evaluated the indication for the CMA testing and the yield of positive tests which included CNVs, candidate gene in AOH areas that could explain the phenotype, and UPD.

Results: CMA tests were performed for 136 patients. The indications included developmental delay, dysmorphic features, growth failure, and multiple congenital anomalies. Consanguinity and AOH areas of more than 4% were seen in 76 (56%). A definite diagnosis was reached based on this test in 30 patient (22%), including 23 patients with CNV (deletion or duplication); 1 patient with UPD (Russell-Silver syndrome); and 6 patients cases were found to have mutations in genes in the AOH area (5 alpha reductase deficiency and alacrima, achalasia, and mental retardation syndrome, Hair-Cartilage syndrome, Deafness autosomal recessive 1A, and Polycystic kidney infantile).

Conclusion: CMA is one of the commonest tests performed in the diagnostic work up of genetic diseases. The yield of CMA can be higher in communities with high consanguinity rates due to the utility of AOH areas, suggesting candidate genes that can explain the phenotype

Biography

Mustafa Alabdullatif has completed his Master's degree and Arab Board of Pediatric from Aleppo University. He worked in different hospitals in KSA, and UAE. Currently, he is working at one of the best tertiary hospital in UAE.

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