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بعض الدراسات الوراثية الخلوية والجزيئية على مرض التوحد

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Abstract : Many cytogenetics studies reported various chromosomal abnormalities associated with autism all over the human genome and it varies from one case to another or from one study to another , also linkage analysis and molecular studies reported over 100 genes to be associated with autism, and based on the literature reviw of the disorder we desided to participate in the genetic research as a start using three approaches; first the cytogenetic approach by constructing karyotypes , second approach was molecular cytogenetic by applying Fluorescence In Situ Hybridization (FISH) technique, and the third approach was on the molecular level using Polymerase Chain Reaction (PCR) technology. Due to difficulties in the process of tissue cultures and karyotyping we decided that it is crucial to run an optimizational experiment before starting with the patients samples especially that these samples are very precious to collect. We have collected 21 blood samples from autistic children of those who undergo for at least 14 charachteristics of those mentioned in the Diagnostic and Statistical Manual of mental disorders (DSM-IV) by the American Psychiatry Association (APA) in 1994 after the signing of a consent form by the guardian of each patient. Tissue cultures were made for all samples and using G-banding technique all samples demonstrate normal karyotypes. Association Study of three chromosomal regions (7q11.23 , 15q11 , 22q11.2) that was mentioned in previous studies for association with autism. FISH was performed according to the probes manufacturer protocol, and results shows no duplications or deletions in all samples, wich indicate that these regions might not be necessarily associated with autism in our samples. There are about 40 single gene disorder that are caused by Trinucleotide Repeat Expansion (TRE) and it is proven that TRE is a general phenomena which is responsible for a growing number of neurological disorders. The behavioral overlap between autism and fragile X syndrome (FXS) which is caused by TRE suggests some overlapping mechanisms. In the last experiment we determined the trinucleotide repeat number (TRN) of CGG in 5 end for the untranslated region of the gene FMR1 which causes FXS and the results show that all samples have a CGG trinucleotide repeat number less than 50 repeats, which means that all children involved in this study have a normal allele for the 5 end for the untranslated region of the gene FMR1