P-705 - COPY NUMBER VARIANTS DISTRIBUTION IN A PAIR OF DISCORDANT MONOZYGOTIC TWINS FOR THE AUTISM SPECTRUM DISORDER

J.Fibla[1], M.Laplana[1], A.Aluja[1], M.Pijuan[2], R.López[2], D.Heine[3]

[1]University of Lleida, [2]Hospital Universitari Arnau de Vilanova, Lleida, [3]Hospital Universitari Son Dureta, Palma de Mallorca, Spain

Introduction: Autism is characterized by a broad spectrum of disorders of communication, language skills, social interaction and behaviour (Autism Spectrum Disorder, ASD). Studies of twins' concordance indicate that 85-90% of the ASD variability can be attributed to a genetic basis with a strong genotype-environment interaction. It has been suggested that copy number variations (CNV) contribute significantly to the phenotypic variability of complex disorders such as autism. 10-15% of monozygotic twin pairs show discordance on ASD.
Objectives: To evaluate CNVs patterns in a pair of male monozygotic twins discordant for ASD.
Aims: To identify ASD candidate genomic regions and genes.
Methods: A pair of discordant monozygotic twins was diagnosed according to ADOS and ADI-R as Autistic Disorder (twin1) and Pervasive Developmental Disorder Not Otherwise Specified (twin2). DNA was obtained from blood and saliva and used to perform a comparative genome hybridization (CGH) analysis with the Agilent 2X400K CGH-array. DNA from each twin was compared with a DNA pool obtained from healthy controls. CGH results were analysed by Agilent Genomic Workbench using ADAM2 algorithm (threshold of 6 and a minimum of 4 probes) to identify CNVs.
Results: X-fragile and Angelman Syndrome aetiology were discarded. We identified common and private CNV in blood and saliva. Two CNV regions (CNVR) located at 16p11.2 and 1p36.13 were identified as private for twin1 and twin2, respectively.
Conclusion: CNVR differ among ASD discordant monozygotic twins. 16p11.2 microdeletion syndrome has been related to autism. An in deep characterization could allow us to identify candidate genes in ASD.

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