PW01-165 - **IDENTIFICATION OF A DE NOVO 22Q11.2 DISTAL DELETION IN AN ADULT FEMALE REFERRED FOR AN ANXIETY DISORDER**

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Velocardiofacial syndrome, nowadays called 22q11 deletion syndrome (22q11DS), involves a common hemizygous interstitial microdeletion at 22q11.2. The syndrome has a highly variable expression including congenital cardiac and vascular anomalies, endocrine dysfunctions, facial dysmorphisms and an increased risk for the occurrence of a specific psychopathological phenotype.

The patient is an 18-years-old female and only child from non consanguineous parents. She was referred because of anxious preoccupations with death. Her history mentioned postnatal feeding problems, surgical correction of septal defects aged 6 months, frequent upper airway infections during childhood, developmental delay and recurrent anxieties. Her somatic phenotype was characterized by nasal speech, minor facial dysmorphisms, bilateral hypoplasia of the thenar eminence. Neuropsychiatric examination demonstrated affective instability, severe anxieties accompanied with social withdrawal, ideas of reference, perseverations and paranoid ideation. Neuropsychological assessment demonstrated executive dysfunctions, attention deficits, low social and interpersonal skills and a disharmonic intelligence profile with limited verbal capacities. Total IQ was 81 indicating low intelligence level. Laboratory analyses and MRI scanning of the brain revealed no abnormalities. A provisional clinical diagnosis of 22q11 deletion syndrome was made. Treatment with 20mg citalopram daily for 2 years resulted in complete remission of anxiety symptoms.

Routine cytogenetic investigation demonstrated a normal 46XX karyotype, and no fragile X. 22q11.2 FISH analysis showed no deletions nor did subsequent MLPA subtelomere deletion testing. SNP array analysis demonstrated a 738,8 kb distal deletion in 22q11.21q11.22.

This report demonstrates the importance of testing for 22q11.2 distal deletions in patients with a mild phenotypical presentation, etiologically suggestive for 22q11DS.

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