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Digeorge Syndrome: Psychotic Risk and ERPS Correlates

A. Mirigliani¹, M. De Lucia¹, C. Mannarelli¹, D. Mannarelli¹, N. Locuratolo¹, A. Buzzanca¹, C. Squarcione¹,

M.C. Torti¹, F. Di Fabio¹, F. Fattapposta¹, M. Biondi¹

¹Department of Neurology and Psichiatry, "Sapienza" Università di Roma, Roma, Italy

The 22q11.2 deletion syndrome is a neurogenetic disorder resulting from a hemizygous deletion. Individuals with 22q11DS present with a wide range of clinical manifestations; an increased risk of behavioral and neurocognitive *sequelae* throughout development have been reported. Approximately 30% of individuals develops a psychotic disorder in adolescence or early adulthood, making this syndrome one of the largest known genetic risk factors for schizophrenia.

The aim of this study is to evaluate some psychophysiological aspects in patients with DiGeorge syndrome in the attempt to recognize earlier specific features able to provide pre-clinic evidence predictive of a possible evolution towards schizophrenia.

Eight subjects with 22q11DS (median age 28,6-29,8±2,3ys), eight psychotic patients and eight matched healthy controls underwent a psychophysiological assessment. CNV and P300 (oddball and Novel paradigm) were recorded. CNV amplitude (total area and two temporal windows, W1 and W2), and P3 parameters were measured.

A total CNV area decrease was found in 22q11DS with respect to psychotic and healthy controls (p=0.04 and p=0.07 respectively). A slight difference was evident at W1 in 22q11DS patients and psychotics vs controls. A N1 latency reduction was observed in 22q11DS patients during Novelty P3 paradigm (p=0.03). Psychophysiological changes in CNV and P3 latency and amplitude have been repeatedly found in schizophrenic patients and interpreted as a deficit in attentional processes.

Data related to our DiGeorge subjects suggest a possible frontal involvement of attentional processes in absence of a psychiatric symptoms. A follow-up study could confirm a predictive role of these ERPs findings in this syndrome