THE COGNITIVE PROFILE OF THE 22Q11.2 DELETION: A UK SAMPLE

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Background: The 22q11.2 deletion is the most frequent microdeletion syndrome. Learning difficulties (LD) are frequently reported with marked variability both within cognitive domains and between subjects. Comorbid psychiatric disorder is common and by early adulthood up to 30% develop a schizophrenia-like psychosis (Murphy et al. 1999; Feinstein et al. 2002; Gothelf et al. 2004a; 2004b). A review of the literature presents the main findings and methodological limitations. Aim: The aim of the study is to explore the cognitive profile in children with the 22q11.2 deletion at Great Ormond Street Hospital in the UK. Method: Thirty one children with the 22q11.2 deletion were examined on standardized tests of intelligence, memory, literacy and numeracy. Results: The mean Full Scale IQ (FSIQ) was 65, in the Mild LD range. Verbal IQ was significantly higher than Performance IQ, with the discrepancy clinically significant in half the sample. Memory function was higher than FSIQ, with no significant differences between verbal and visual memory. Memory was significantly higher for single than complex verbal information. Verbal role learning was a strength. Basic reading was significantly higher than mathematics, especially in those with FSIQ below 70. All other results remained significant in children with an FSIQ above 70 and when controlling for age, gender and curdiae surgery. Conclusions: Children with the 22q11.2 deletion in this sample have LD with a specific but heterogeneous cognitive profile. Heterogeneity is contributed to by many factors including presence of schizophrenia, genotype, nature of microdeletion, developmental effects and cardiac defects. Implications for schizophrenia research are reviewed. This study replicates the findings of previous research, apart from the weakness of visual memory compared to verbal memory.

The present study indicates that the cognitive profile of 22q11.2 deletion is heterogeneous and influenced by factors such as age, gender, and other medical conditions. It highlights the need for individualized assessment and support for these children.

NEUROTROPHIC FACTORS AND SCHIZOPHRENIA: RELATION TO COGNITION

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Gial cell line-derived neurotrophic factor (BDNF) and Brain derived neurotrophic factor (BDNF) may play an important role in various neurodevelopmental processes which are assumed to be abnormal in schizophrenia. The objective of this study was to determine the relationship between peripheral BDNF and GDNF serum and gene expression levels and cognition in schizophrenia. Thirty five patients diagnosed with schizophrenia according to DSM-IV (APA, 1994) and 17 healthy volunteers were included in the study. The patients were recruited from inpatient and outpatient departments of Psychiatry of Dokuz Eylul University Hospital. Patient and control groups were comparable for gender and years of education but not for age. Total RNA extracted from lymphocytes of individuals was amplified by RT-PCR. Quantitative real time PCR using SYBR Green I was used to quantify the expression of BDNF and GDNF genes. Relative expressions of BDNF and GDNF were normalized with beta-actin as housekeeping gene. The Delta-DeltaCt method was used for the analysis of relative expression. Serum BDNF and GDNF levels were measured by sandwich ELISA. Neuropsychological tests were administered to evaluate attention, executive functions, verbal and visual learning and memory, working memory, verbal fluency and motor function. The relationship between peripheral BDNF and GDNF serum and gene expression levels and measures of cognitive tests were examined with Pearson correlation test. Serum BDNF level was related to a measure of verbal learning and GDNF gene expressions levels were correlated to measures of verbal learning and memory, executive function and motor function in healthy subjects. Patient group revealed a negative correlation between Serum BDNF level and a measure of motor function (r = -0.35) and no other correlations were found between serum and gene expression levels of BDNF-GDNF and measures of neuropsychological tests in patients with schizophrenia. There are a few studies indicating a possible relation between BDNF and cognitive functions particularly learning-memory and executive functions in healthy subjects. The number of studies investigating the relationship between cognition and neurotrophic factors in schizophrenia are a few and their results are conflicting. This study examined this relationship using a wide cognitive battery and did not find a significant relationship.

VERBAL AND VISUAL THEORY OF MIND IN A SCHIZOPHRENIC AND BIPOLAR POPULATION

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Theory of mind (ToM) is described as the mental capacity to assign emotions, thoughts, or feelings to others. It has been hypothesized that this trait plays an integral role in social functioning and interpersonal relations. This study inquired into the existence of a deficit in ToM in schizophrenic (SZ) and bipolar populations (BPD), and whether this deficit is related to the level of social functioning, symptom severity, and/or intellect. The tests used included a verbal task, the Hinting Task, and a visual task, the Eyes Test, both originally established for autistic adult populations. This study aims to not only investigate group differences, but also to investigate the possible strategic differences in ToM. SZ, BPD, and demographically matched healthy participants (CO) were given the Intelligence Quotient (IQ), Social Functioning Scale (SFS), the Hinting Task, the Eyes Test and a symptoms rating scale. In the Eyes Test, the participants completed a computer task in which they were asked to choose the appropriate emotion to describe the eyes presented from a list of four adjectives; all term definitions are provided. The Hinting Task is administered as an interview where ten scenes are described where one of the actors is hinting at an action. The subject is asked what the actor is hinting at based on the description. These scores were compared to a series of empathy, social functioning, and intelligence tests scores. SZ showed a deficit when compared to the CO, while BPD was not significantly different from either group. Eyes test scores over all groups were positively correlated with the Social Functioning Scale, as well as with IQ scores. The symptoms ratings scale showed negative trend between positive symptoms and Eyes Test Score in SZ. Overall Hinting Task scores are not significantly correlated with the Eyes Test scores. Hinting Task is only positively correlated to the Verbal IQ and one subscale of SFS. There was a trend toward group differences in the Hinting Task, but BPD did not differ significantly from CO or SZ. This suggests that there is the possibility of a socially related deficit in ToM with...