W01-02 - NEUROPSYCHIATRIC ASPECTS OF THE 22Q11 MICRODELETION SYNDROME

M.Gawlik, B.Pfuhlmann, G.Stöber

Department of Psychiatry, Psychosomatics and Psychotherapy, University of Würzburg, Würzburg, Germany

Introduction: 22q11 microdeletion is the most common genetic risk factor for psychoses with an assumed prevalence of 1:2000 to 1:4000. Majority of deletions are sporadic encompassing ~ 3 million base pairs with 40 genes. Up to one third of the subjects with 22q11 deletion syndrome develop psychotic symptoms. It is still on debate whether these patients show a clinical subtype of schizophrenia or organic psychotic and organic personality syndromes.
Objectives: To discuss the role of 22q11 microdeletion syndrome in psychoses.
Aims: To focus on neuropsychiatric aspects of the 22q11 microdeletion syndrome.
Methods: Review and comment on neuropsychiatric findings in 22q11 microdeletion syndrome.
Results: The cognitive performance of subjects with 22q11 microdeletion is characterized by mild intellectual disability with strengths in verbal functioning and relative weaknesses in the areas of visuospatial memory and executive functioning. The frontostriatal and frontoparietal neural networks are thought to be particularly affected. Psychiatric phenotype shows a great variability. Around 30% of patients with 22q11 deletion syndrome develop paranoid syndromes but without negative symptoms or a defective state. Other clinical pictures are attention-deficit/hyperactivity disorder, bipolar depression or autistic spectrum disorders. Structural brain studies reported a decrease of brain volume with pronounced changes in the prefrontal cortex, parietal lobe, cerebellum, and temporal lobe. Candidate genes locating within 22q11 deletion could effect brain development and neuropsychiatric outcome in patients, but evidence is still far from being conclusive.
Conclusion: The role of 22q11 microdeletion in psychoses is still not defined; phenotype correlation could be helpful to elucidate biological bases.

https://doi.org/10.1016/S0924-9338(12)75669-9 Published online by Cambridge University Press