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META-ANALYSIS OF MAGNETIC RESONANCE IMAGING STUDIES IN VELOCARDIOFACIAL SYNDROME (VCFS)

G. Tan[1], D. Arnone[2], A.M. McIntosh[3], K.P. Ebmeier[4]

[1]Section of Brain Maturation, Department of Psychological Medicine, Institute of Psychiatry, London, [2]Neuroscience and Psychiatry Unit, University of Manchester, Manchester, [3]Royal Edinburgh Hospital, University of Edinburgh, Edinburgh, [4]Department of Psychiatry, University of Oxford, Oxford, UK

Introduction: Velocardiofacial syndrome (VCFS) is a common genetic disorder due to a micro deletion on chromosome 22q11. This region includes several risk-associated genetic variants, including COMT, and VCFS is associated with a substantially increased risk for schizophrenia. As such, VCFS may serve as a valuable model for clarifying the neuroanatomical changes associated with genetic risk for psychosis.

Methods: A systematic literature search was conducted. Studies were included if they presented original data and were published by March 2008, compared subjects with VCFS and healthy controls and reported measures of brain regions according to SI units as mean and standard deviation. Data extracted from the studies included diagnosis, demographic variables and IQ. Statistical analysis was conducted using STATA 8.0 supplemented by 'Metan' software.

Results: Twenty studies were retrieved. All measures were expressed in volumes apart from the corpus callosum (area). Subjects with VCFS showed reduced total brain volume (N=156 versus N=138), ([ES]=1.04, 95% CI:1.40, -0.67), with no significant heterogeneity or publication bias. This reduction was reflected in total hemisphere grey and white matter. Prefrontal, parieto-occipital and temporal cortices appeared to be particularly affected. A number of sub-cortical areas also showed decreased volumes including the hippocampus and putamen. In contrast, callosal areas were increased in VCFS.

Conclusion: In relation to controls, subjects with VCFS present with an overall reduction in brain volumes and specific abnormalities in multiple cortical and subcortical brain regions. These abnormalities may explain partly why VCFS is associated with a greatly increased risk of psychosis and other psychiatric disorders.

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