The Importance of Research on the Relatives of Schizophrenia Patients – Towards Endophenotypic Markers

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The endophenotype concept of schizophrenia represents an important approach in the exploration of the neurobiology of the illness. Gottesman and Shields (2003) described an endophenotype as an internal, intermediate phenotype (i.e., not obvious to the unaided eve) that fills the gap between genes and diseases. Endophenotypes should be: (1) associated with the illness in the population, (2) heritable, (3) state-independent, (4) found in unaffected family members at a higher rate than in the general population, and (5) shown to cosegregate with the illness within families. As we can see an important characteristic of an endophenotype, that it can be found among the healthy, first-degree relatives of patients with schizophrenia. Minor physical anomalies (MPAs) and social cognition (SG) are suggested as endophenotypes on account of the findings that MPAs and SG deficits are more common in schizophrenia patients than in healthy controls, which can be found also in remission and in few studies higher prevalences were found in healthy first-degree relatives (Xu et al., 2011; Bora & Pantelis, 2013).

Recently we evaluated two systematic reviews of studies on these two potential endophenotypes (MPAs and SG) to confirm the possibility of them as biological and cognitive markers of the illness. We planned to explore data on MPAs and SG among the relatives of schizophrenia patients. We evaluated two researches of studies published in PubMed, Medline,Web of Science and PsychINFO between the period of 1968 and 2014 (MPAs) and 1980 and 2014 (SG). 11 studies on the appearence of minor physical anomalies in the relatives of schizophrenia patients were found with mixed results, while 15 studies and two meta-analyses were analysed on social cognition studies among the first-degree relatives of patients with schizophrenia, the exploration of the latter studies showed also diverse findings (Hajnal et al., 2014; Tényi et al., 2014). We confirm that further research is needed to clarify social cognition and minor physical anomaly alternations as endophenotypic markers of schizophrenia.

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