AS23-02 - HOW IMPORTANT ARE RARE GENETIC VARIANTS IN PSYCHIATRIC DISORDERS? EXPERIENCES FROM SCHIZOPHRENIA, AUTISM, AND BIPOLAR DISORDER RESEARCH

D.Rujescu

Dept. of Psychiatry, University of Munich (LMU), Munich, Germany

There is a long lasting assumption in psychiatric genetics that common genetic variants with small effects are enhancing the risk to develop e.g. schizophrenia. Although debated for some time, the other side of the coin namely that rare genetic variations with large effects may account for a significant number of schizophrenia cases has been somehow neglected. However, rare variants with large effects on schizophrenia are known for a long time. One represents a private translocation in a Scottish family which disrupts DISC1. A more common deletion of chromosome 22q11 has also been repeatedly reported to substantially enhance the risk for developing schizophrenia. Structural chromosomal abnormalities are emerging as an important genomic cause of neuropsychiatric diseases, including mental retardation, autism and more recently schizophrenia. Did these new studies investigate only the tip of an ice berg? Are a substantial number of schizophrenia cases caused by rare copy number variations? It is also interesting that some of the genes may show association not only to schizophrenia but also to mental retardation and autism, an example is neurexin 1 or deletions at 1q21.1, 15q11.2 and 15q13.3. Are we going to define a number of new diseases at the interface between mental retardation.

https://doi.org/10.1016/S0924-9338(12)74017-8 Published online by Cambridge University Press